NOTES
INFLAMMATORY ARTHRITIS

GENERALLY, WHAT IS IT?

PATHOLOGY & CAUSES

- Musculoskeletal disease subset; known immune component underlying disease
- Underlying trigger/cause not always understood

SIGNS & SYMPTOMS

- Painful, warm, stiff joints
- Variable extra-articular symptoms

DIAGNOSIS

LAB RESULTS

Synovial fluid analysis

- Cloudy yellow appearance
- White blood cell count (WBC) > 5,000
- Polymorphonuclear neutrophils (PMNs) < 25%

TREATMENT

MEDICATION

Anti-inflammatory medication

- Common NSAIDs
- Immunologically-targeted therapy
  - Anti-cytokine therapy (e.g. adalimumab)

ANKYLOSING SPONDYLITIS

osms.it/ankylosing-spondylitis

PATHOLOGY & CAUSES

- *Group:* seronegative spondyloarthritides
- *Characteristics:* articular cartilage destruction, bony joint fusion (ankylosis) → primarily spine, sacroiliac joints
- AKA rheumatoid spondylitis, Marie–Strümpell disease
- Autoimmune self-reactivity believed to underlie pathophysiology
  - Strong HLA-B27 association (MHC I serotype; positive in 90% of affected individuals)
  - *Relative risk for HLA-B27 individuals:* 100-200x
  - IL-23 receptor gene also implicated
  - Abnormal IL-23 cytokine regulation → naive CD4+ T cell → self-reactive Th17 cells
- Associated with Crohn's disease, ulcerative colitis

RISK FACTORS

- Biological sex
  - 3x ↑ individuals who are biologically male

COMPLICATIONS

- Aortic regurgitation
  - Aortic aneurysm → aortic valve annulus stretched → regurgitation
- Uveitis
- Enthesitis (tendinous insertion inflammation)
- Dactylitis ("sausage fingers")
- Decreased pulmonary function
- Thoracic-rib articulation spondylosis → ↓ chest wall expansion across respiratory cycle
- Secondary amyloidosis

SIGNS & SYMPTOMS

- Symptoms develop teens-20s
  - Lower back pain, spinal immobility
- Peripheral large joints (hips, knees, shoulders) involved in ⅓ of individuals
- Morning stiffness; improves throughout day, with exercise
- Untreated disease → extenuated kyphosis of spine

DIAGNOSIS

DIAGNOSTIC IMAGING

Lumbar spine radiograph

- Diagnostic → sacroileitis
- Progression of early findings
  - Subchondral erosions (pseudo-widening effect on X-ray) → sclerosis → sacroiliac joint fusion
- Late findings (10+ years of disease)
  - "Bamboo spine": prominent syndesmophytes (bony growth inside ligaments), diffuse calcification of paraspinal ligaments, spinal osteoporosis

LAB RESULTS

- ↑ erythrocyte sedimentation rate (ESR), C-reactive protein (CRP) suggestive, not diagnostic
- ⊖ Rheumatoid factor (RF)

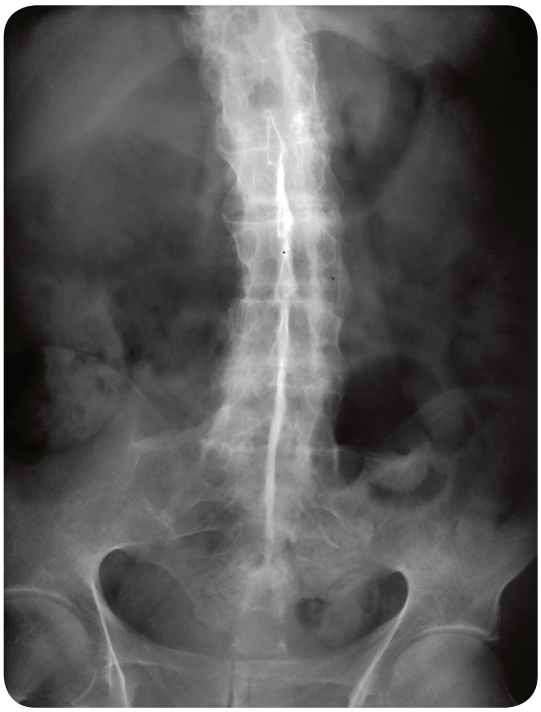

Figure 111.1 An X-ray image of demonstrating bamboo spine and the dagger sign in an individual with ankylosing spondylitis.

OTHER DIAGNOSTICS

- Family history
- Physical examination
  - ↓ Spine flexion/extension and ↓ lateral range of motion

TREATMENT

MEDICATIONS

- NSAIDs (maximum daily dosing recommended)
  - First line for pain, stiffness
- TNF-alpha inhibitors
  - *Etanercept*: fusion protein (IgG1 Fc region, TNF alpha receptor); intercepts circulating TNF-alpha, competes with body's TNF alpha receptors
  - *Infliximab, adalimumab, certolizumab*: anti-TNF alpha monoclonal antibodies
  - *Sulfasalazine*: if TNF-alpha therapy ineffective; recommended for peripheral joint disease

Figure 111.2 The skeleton of an individual with ankylosing spondylitis. The lumbar and cervical spine have ossified completely and become fused.

OTHER INTERVENTIONS

- Exercise therapy
  - Home exercise therapy/formal physical therapy regimens
- Tobacco use cessation
- Heat, ice packs

GOUT

osms.it/gout

PATHOLOGY & CAUSES

- Episodic, arthritic disorder
- Monosodium urate crystallization in, around joint spaces; when left untreated, can manifest as tophi in chronic arthritic disorder
- *Monosodium urate (MSU):* purine and pyrimidine (nitrogen containing heterocycles; DNA components) → primary sources of uric acid; released when cells broken down
  - Limited solubility in plasma (only 6.8mg/dL)
  - Poorer solubility in joint space → lower temperature, synovial fluid composition favor precipitation
  - Sources of nidus (precipitates crystals) include collagen fibers, chondroitin sulfate, proteoglycans, cartilage fragments
  - Physiologic pH of 7.4 → uric acid loses cation, adds Na^+ → MSU crystals
- *Damage pathway:* MSU precipitate into joints → complement cascade activated, cytokines produced → leukocyte recruitment → macrophages phagocytose MSU → inflammasome activates caspase-1 → produce IL-1 and other proinflammatory cytokines → ↑ ↑ ↑ leukocyte recruitment, cytokine production
- Classification
  - 90% primary/idiopathic
  - 10% secondary

CAUSES

- ↑ production of uric acid, purines (most common)
- Diet high in red meat, shellfish, anchovies, organ meat
- ↑ cell turnover
- Cancer treatment → tumor lysis syndrome
- Polycythemia vera (5–10% develop gout)
- Lesch-Nyhan syndrome
  - Hypoxanthine guanine phosphoribosyl transferase (HGPRT) deficiency interrupts purine salvage pathway → ↑ degradation of purines → ↑ uric acid production
- Dehydration, alcoholic beverage consumption → ↓ clearance of uric acid
- Chronic kidney disease

RISK FACTORS

- Age
  - 20–30+ years of hyperuricemia → ↑ risk
- Biological sex (↑ individuals who are biologically male)
- Genetic
  - *HGPRT* (X-linked); *URAT1*, *GLUT9* (both involved in urate transport/homeostasis)
- Heavy alcohol consumption
- Obesity
- Drugs that ↓ urate excretion/↑ production (e.g. thiazides, aspirin)
- Glucose metabolization abnormalities (e.g. diabetes mellitus)
- Chronic lead toxicity → saturnine gout
  - Most common risk factor in U.S. is moonshine consumption → lead-lined stills

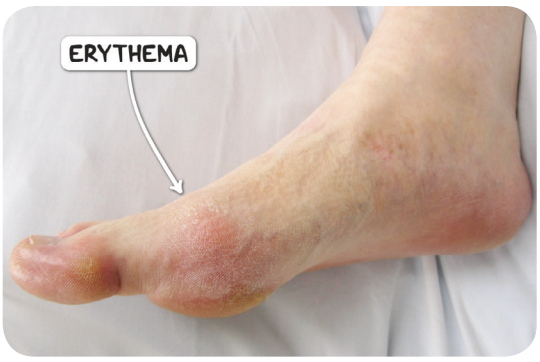

Figure 111.4 Gout of the great toe presenting as erythema of the overlying skin.

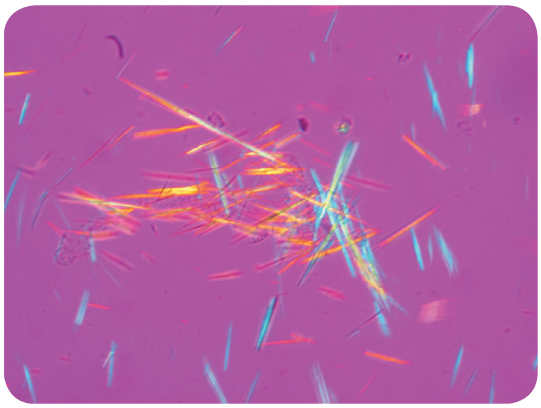

Figure 111.3 Urate crystals will display negative birefringence on polarised light microscopy.

COMPLICATIONS

- Gravel/stone passage → renal colic
- Renal failure → death in 20% individuals with chronic gout
- ↓ quality of life, generally not lifespan

SIGNS & SYMPTOMS

Acute, episodic arthritis

- Nocturnal onset
  - Awakening with complaints, e.g. "feeling like toe on fire"
- Most severe pain remits within first hours; pain can last days–weeks
- Painful, warm, erythematous, and swollen joint → ↓ range of motion → disability

First episode

- Commonly monoarticular; 50% of cases include first metatarsal joint (aka podagra)
- Asymptomatic period months–years → subsequent episodes (mono- or polyarticular)
  - 90% of other joints involved, progressively (ankle > heels > knees > wrists > fingers > elbows)
  - ↑ episodes, polyarticular effects without treatment

Chronic disease (tophaceous gout)

- On average, around 12 years after initial attack
- MSU deposition (joint spaces/affected cartilage)
- Painless, pedunculated mass; palpitation may discolor overlying skin
- Joint's range of motion sometimes limited
- Kidney complications take one of two forms
  - Symptoms of colicky flank pain, hematuria → uric acid nephrolithiasis
  - ↓ urine output, difficulty voiding → urate nephropathy

DIAGNOSIS

DIAGNOSTIC IMAGING

Radiographic/ultrasound/CT scan

- Joint destruction, bony erosions (rarely present on the first acute episode)
- Imaging findings become more likely with disease duration

X-ray

- Radiolucent uric acid nephrolithiasis

LAB RESULTS

Synovial fluid analysis

- MSUs in context of acute, arthritic episodes

Polarized light microscope

- Long, slender needle-shaped crystals in synovial fluid, neutrophil cytoplasm
  - Negatively birefringent; yellow under parallel light, blue under perpendicular light

OTHER DIAGNOSTICS

- Histological analysis of chronic tophaceous arthritis
  - Large aggregations of MSU surrounded by inflammatory reaction of foreign body giant cells
  - Hyperplastic, fibrotic, thickened synovium → pannus formation → destruction of underlying cartilage, juxta-articular bony erosions

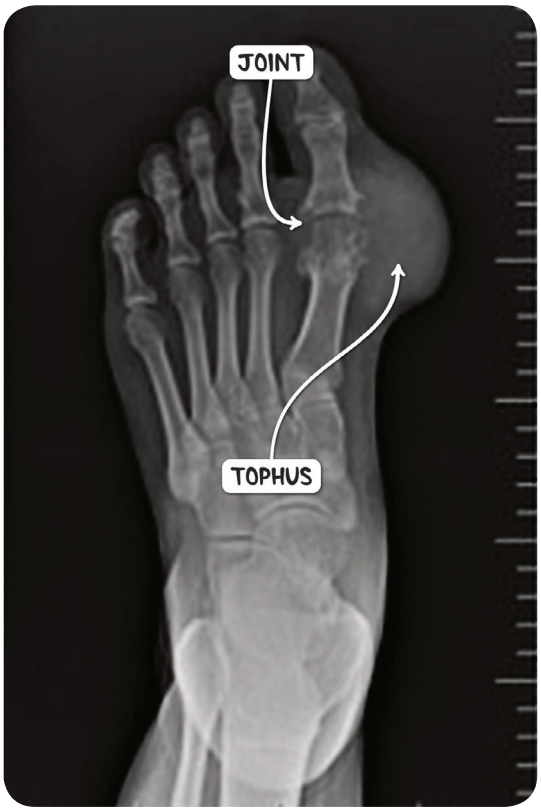

Figure 111.5 An X-ray image of the foot showing destruction of the first metatarsophalangeal joint by arthritis secondary to gout. There is an overlying gouty tophus.

- Histological analysis of gouty nephropathy
  - MSU (with/without tophi) deposits in medullary interstitium/tubules

TREATMENT

MEDICATIONS

Acute flare therapy

- Anti-inflammatory treatment ASAP within acute flare → rapid, complete resolution faster
- Glucocorticoids (oral and/or intra-articular injections)
- NSAIDs (i.e. naproxen, indomethacin)
- Colchicine (inhibits leukocyte migration)
- Biologic agents (IL-1 inhibitors)

Management and prevention

- Limit medications that alter urate balance (e.g. thiazides, aspirin)
- Initiate medications that ↓ uric acid levels
- Xanthine oxidase (XO) inhibitors (allopurinol, febuxostat)
  - *Mechanism of action:* directly inhibits enzyme → urate production, stimulates purine base reutilization → ↓ ↓ ↓ urate concentration
- Uricosuric medications (probenecid)
  - ↑ urate excretion at kidney
- Uricase medications (rasburicase)
  - Mimic enzyme that catalyzes urate conversion → allantoin (more soluble purine degradation product); enzyme absent in humans

OTHER INTERVENTIONS

- Benefit largely due to ↓ development/worsening of obesity, cardiovascular disease, diabetes mellitus
- Diet modification
  - Limit/avoid soda, red meat, seafood
- Alcohol moderation
- ↑ physical activity

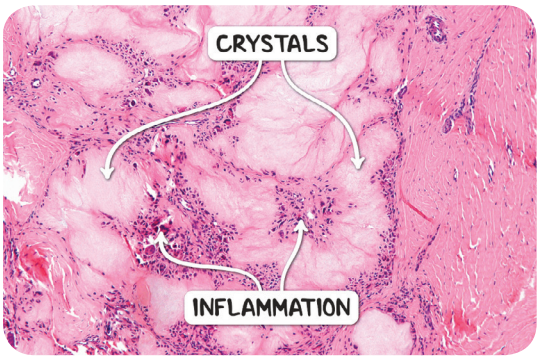

Figure 111.6 The histological appearance of a gouty tophus. There is a large aggregate of urate crystals which is associated with granulomatous inflammation.

JUVENILE IDIOPATHIC ARTHRITIS

osms.it/juvenile-idiopathic-arthritis

PATHOLOGY & CAUSES

- Arthritic symptoms of unknown etiology; present < 16 years old for ≥ six weeks
- Unknown pathophysiology; appears related to TH1 and TH17 cells → cell mediators
  - IL-1, IL-17, TNF-gamma

RISK FACTORS

- HLA and PTPN22 variants

COMPLICATIONS

- 10% develop disability in adulthood

SIGNS & SYMPTOMS

- Arthritis
  - Oligo- or polyarticular involvement; large joints affected > small
- Rheumatoid nodules, factor usually absent

DIAGNOSIS

LAB RESULTS

- Antinuclear antibodies may be ⊕ or ⊖

TREATMENT

- Similar to rheumatoid arthritis
- Some success with IL-6 R antibody biologic disease-modifying antirheumatic drug (DMARD)

PSEUDOGOUT

osms.it/pseudogout

PATHOLOGY & CAUSES

- Calcium pyrophosphate (CPP) crystal depositions in articular cartilage
- AKA chondrocalcinosis
- Unlike gout, hyperuricemia
  - No known direct substance concentration → ↑ crystal formation
- Pathway of joint inflammation, destruction similar to gout
  - Articular cartilage proteoglycans degraded, serve as nidus for crystal formation around chondrocytes
  - CPP crystals precipitate around chondrocytes → complement cascade activated, cytokines produced → leukocyte recruitment → macrophages phagocytose MSU (inflammasome activates caspase-1) → produce IL-1, other proinflammatory cytokines → further leukocyte recruitment, cytokine production

CAUSES

- Sporadic (idiopathic)
- Hereditary
  - Autosomal dominant version → early manifestation, more severe symptoms
  - Also associated with osteoarthritis
  - Mutations in pyrophosphate transport channel
- Secondary to previous joint damage, hyperparathyroidism, hemochromatosis, hypothyroidism, ochronosis, diabetes

RISK FACTORS

- Age
  - Usually affects individuals > 50 years; by > 85 years → 30–60% prevalence
- ↓ magnesium levels

COMPLICATIONS

- Significant joint damage
  - ≤ 50% of individuals

SIGNS & SYMPTOMS

- Episodic joint pain
  - Knee most commonly affected; followed by wrists → elbows → shoulders → ankles
- Duration of several days–weeks
- Oligo- or polyarticular
- Frequently asymptomatic

DIAGNOSIS

OTHER DIAGNOSTICS

Histological analysis

- Gross
  - Chalky, white, friable
- Microscopic
  - Aggregates stain as blue/purple, oval
- Crystals
  - Rhomboid, ⊕ birefringent
- Crystallization first develops in articular cartilage, menisci, intervertebral discs

Physical examination

- Acutely painful, inflamed joint; commonly knee

TREATMENT

MEDICATIONS

Acute flares

- NSAIDs
- Colchicine
- Glucocorticoids

Management and prevention

- Colchicine

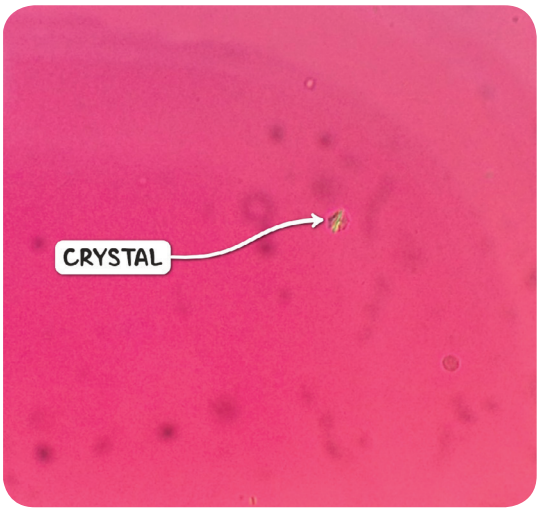

Figure 111.7 A calcium pyrophospahte crystal in joint fluid aspirated from the knee of an individual with pseudogout.

OTHER INTERVENTIONS

- Treat underlying disorder (if known)
- Symptomatic therapy similar to gout treatment

GOUT VS. PSEUDOGOUT

	GOUT	PSEUDOGOUT
CRYSTALS	Monosodium urate (MSU)	Calcium pyrophosphate (CPP)
CRYSTAL HISTOLOGY	Needle-shaped; ⊖birefringent	Rhomboid; ⊕birefringent
AGE OF ONSET (YEARS)	May be young, largely RF*-dependent	50+
MOST COMMONLY AFFECTED JOINT	1st MTP* joint	Knee

*RF - risk factor, MTP - metatarsophalangeal joint

PSORIATIC ARTHRITIS

osms.it/psoriatic-arthritis

PATHOLOGY & CAUSES

- *Group:* seronegative spondyloarthritides
- Associated with psoriasis
- Affects peripheral, axial joints; ligaments, tendons (entheses)
- Abnormal T cell response to unknown culprit antigen
  - T_H1, T_H17 cells thought responsible → stimulate activated $CD8^+$ T cells → cytokine, growth factor environment change/destroy local tissue
  - Implicated synovial cell mediators: IL-1, IL-6, TNF-alpha, IL-8
  - Synovial fibroblasts interact with immune response → secreting IL-1beta, IL-6, and platelet-derived growth factors (PDGF)
  - Affected synovia marked with increased vascularity → ↑ leukocytic entryways

TYPES

- *Mild:* one joint involved/responds to NSAIDs
- *Moderate-severe:* NSAID-resistant
- *Severe:* polyarticular, erosive; functional limitation

CAUSES

- Local trauma induces dysregulated immune response → local tissue destruction
  - AKA Koebner phenomenon
- 10% of psoriatic individuals develop arthritis symptoms

RISK FACTORS

- HLA-B27
- HLA-Cw6
- Obesity
- Associated diseases
  - Myopathy, enteropathy, AIDS
- Age: 30 50 years old

SIGNS & SYMPTOMS

- Predominantly peripheral arthritis of hands, feet
- Distal interphalangeal (DIP) joint first affected, asymmetrically distributed in > 50% of individuals
- Sacroiliac joint affected in 20% of individuals
- Degree of joint involvement may be mild/progress → severe, disfiguring disease as in rheumatoid arthritis (RA)

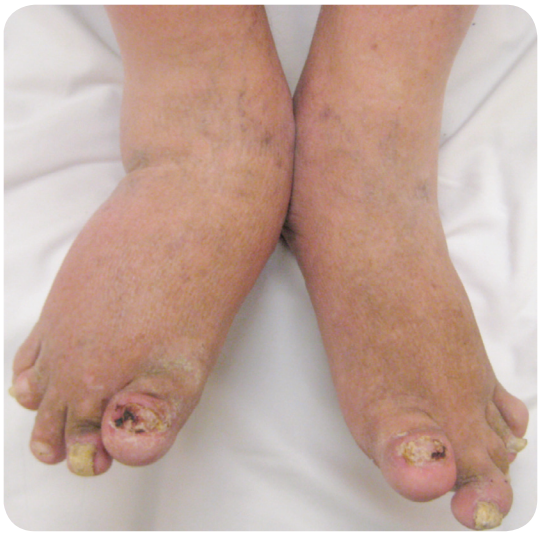

Figure 111.8 The feet of an individual with psoriatic arthritis. There is inflammation of the ankle and the interphalangeal joints as well as psoriatic nail changes.

DIAGNOSIS

DIAGNOSTIC IMAGING

- Characteristic "pencil-in-cup" malformation at DIP joint

OTHER DIAGNOSTICS

Histological analysis

- Similar to RA, but symptoms not as severe, remissions more frequent, joint destruction less frequent

History

- 40% of affected individuals have first-degree relative with psoriatic arthritis

Physical examination

- Integument examination consistent with psoriasis
- Papules, plaques with silver scales on extensor surfaces (fingers, knees, elbows)
- Commonly affects scalp, nails ("nail pitting")
- Musculoskeletal examination consistent with arthritis
- Asymmetric involvement of both peripheral, axial joints
- Commonly affects DIP joints under skin manifestations

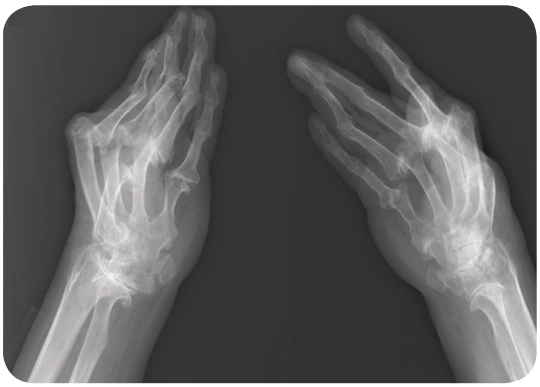

Figure 111.9 An X-ray image of the hands of an individual with long-standing psoriatic arthritis which has progressed to arthritis mutilans. Telescoping of the phalangeal joints is visible.

TREATMENT

MEDICATIONS

Mild disease

- NSAIDs

Moderate-severe disease

- Conventional (DMARD) therapy
- Methotrexate (MTX; co-treat with daily folic acid), leflunomide (does not also target skin disease)
  - Both require eliminating alcohol intake

Severe disease

- 'Biologic' DMARD (e.g. TNF inhibitor)
- Etanercept, adalimumab, infliximab, certolizumab
  - All require latent TB screening before initiation therapy initiation
- Anti-IL-17 'biologic' (e.g. secukinumab, ixekizumab, brodalumab)

OTHER INTERVENTIONS

- Exercise, physical therapy, occupational therapy
- Weight reduction

REACTIVE ARTHRITIS

osms.it/reactive-arthritis

PATHOLOGY & CAUSES

- *Group:* seronegative spondyloarthritides
- Characterized by triad
  - Arthritis, nongonococcal urethritis/cervicitis, conjunctivitis
- AKA Reiter syndrome
- *Hypothesis:* autoimmune reaction to prior infection of GU/GI system
- *Genitourinary triggers:* urethritis/cervicitis
  - *Common pathogen:* *Chlamydia trachomatis*
- *Gastrointestinal triggers:* diarrheal illness
  - *Common pathogens:* *Shigella*, *Salmonella paratyphi*, *Yersinia enterocolitica*, *Campylobacter jejuni*
- RF
- HIV ⊕
- HLA-B27 ⊕ (80+% of affected individuals)

COMPLICATIONS

- Digital tendon sheath synovitis → dactylitis ("sausage" finger/toe)
- Tendoligamentous insertion sites ossification → calcaneal spurs, bony outgrowths
- Severe spinal disease; becomes indistinguishable from ankylosing spondylitis
- Extra-articular involvement
  - Inflammatory balanitis, conjunctivitis, cardiac conduction abnormalities, aortic regurgitation

SIGNS & SYMPTOMS

Arthritic symptoms

- Develop several weeks post-initial infection
- Common, early symptoms
  - Joint stiffness, low back pain
- Days later
  - Painful joints, effusion, lack of mobility
- Most asymmetrically affected joints
  - Ankles, knees, feet; upper extremity involvement less common

Other symptoms

- Fever, malaise, weight loss, fatigue
- Symptoms' severity waxes, wanes; usually lasts 1.5–6 months
- Recurrent arthritic episodes, tendonitis, lumbosacral pain in 50% of individuals
- Keratoderma blennorhagicum
  - Vesiculopustular, waxy lesions on the soles or palms

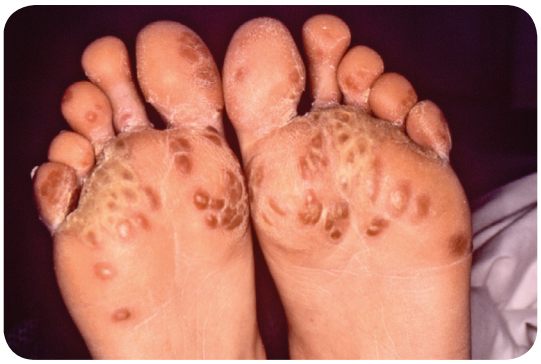

Figure 111.10 Keratoderma blennorhagicum on the feet of an individual with reactive arthritis.

DIAGNOSIS

DIAGNOSTIC IMAGING

X-ray

- Involved joint
  - No specific diagnostic changes
- Negative for stress fractures, other forms of arthritis

LAB RESULTS

Synovial fluid analysis

- Absence of joint space infection, crystals

Cultures

- May be helpful if GI/GU symptoms ongoing → identify well-associated bacteria

OTHER DIAGNOSTICS

History

- Preceding illness, rapid-onset arthritis/systemic symptoms
- Arthritic presentation often too late for stool/urine culture (for GU/GI trigger)

Physical examination

- Lower extremity joint involvement as above

TREATMENT

MEDICATIONS

- NSAIDs
- Glucocorticoids; intra-articular, systemic formulation available
- Resistant/chronic (> six months) disease
  - DMARD (e.g. sulfasalazine, MTX, azathioprine)
- Antibiotics not recommended
  - *Exception:* triggering disease process (GI/GU diarrhea/urethritis/cervicitis) ongoing
- Skin involvement (if present) → topical salicylates

OTHER INTERVENTIONS

- Conjunctivitis (if present) → ophthalmology referral

RHEUMATOID ARTHRITIS

osms.it/rheumatoid-arthritis

PATHOLOGY & CAUSES

- Systemic, chronic, autoimmune inflammatory disorder involving joint synovium
- May progress to disfigurement → cartilaginous, bony damage over time
- Dual hit hypothesis (genetics and environment)
  - *Genetics:* HLA-DR1 or DR4 genetic predisposition → thought to underlie the immune pathogenesis pathway below
  - *Environment:* cigarette smoke, pathogen (i.e. gut bacteria) → may contribute to unknown 'arthrogenic agent', trigger immune response

PATHOLOGY

- *$CD4^+$ T cells:* react with arthrogenic agent (unknown; thought to be a microbe/self-antigen) → cytokine production → IFN-gamma (T_H1 product) → activate macrophages and synovial cells → synovial, immune cell proliferation → swollen synovial tissue (also known as pannus formation) → IL-17 (T_H17 product) → recruit neutrophils and monocytes → TNF-alpha and IL-1 (macrophage product) → stimulate synovial cells → protease release → hyaline cartilage destruction → ↓ cartilaginous buffer → bone on bone articulation → ↑ bone destruction
- *RANKL (on T cells):* activate osteoclasts' RANK receptor → bone resorption

- *Synovial cells:* directly responsible for protease release, contribution to cytokine milieu
  - Germinal centers within synovium include plasma cells → antibodies against self-antigens, i.e. autoantibodies → specific for citrullinated peptides (CCPs)/arginine residues converted to citrulline
  - Antibodies against fibrinogen, type II collagen, alpha-enolase, vimentin → form antibody-antigen complexes → deposit into joints
  - Antibodies (usually IgM or IgA) against Fc regions of IgG antibodies form RF → deposit into joints
- Chronic inflammation → angiogenesis → increase inflammatory cell response → further joint involvement

Extra-articular involvement

- Pyogens (i.e. IL-1)
  - → Hypothalamus → fever
- Skeletal
  - Protein breakdown
- Skin
  - Macrophage and lymphocytes recruitment → cycle of activation/recruitment → cells around a central necrotic mass → rheumatoid nodules
- Blood vessels
  - ↑ cytokines and ↑ circulating immune cells → altered endothelial cells → ↑ atheromatous plaques formation
- Liver
  - Under chronic inflammation → ↑ hepcidin production → ↓ iron absorption → anemia
- Lung
  - ↑ fibroblasts → lung fibrosis (AKA Caplan syndrome) → ↓ gas exchange (+/- pleural effusion)

COMPLICATIONS

Autoimmune

- AA amyloidosis
- Sjögren syndrome
- Scleritis

Cardiovascular

- ↑ Atheromatous formation → ↑ MI, CVA risk
- Pericarditis → ↑ pericardial effusion risk

Hematologic

- Anemia
- Felty syndrome (RA, splenomegaly, neutropenia)

Musculoskeletal

- Rheumatoid nodules
  - Can form in any body tissue
- Baker (popliteal) cyst formation

Neurological

- Carpal tunnel syndrome
- Mononeuritis multiplex
- C1-C2 instability → ↑ risk of subluxation → spinal cord impingement risk → neurologic involvement
- Serious complication if unknown at time of intubation

Pulmonary

- Pleuritis → ↑ risk of pleural effusion (characteristically ↓ glucose, ↓ complement)
- Interstitial lung disease
- Caplan syndrome

SIGNS & SYMPTOMS

Inflammatory polyarthritis

- Commonly symmetrically affects multiple (> five) joints
- First smaller joints - MCP, PIP, MTP
- Avoids DIP joint
- Chronic disease → ↑ larger joint involvement

Joint characteristics

- Warm, red, and painful joints
- Morning stiffness (lasting > one hour)

Malformation

- Ulnar deviation of MCP joints
- Boutonniere (buttonhole) malformation
- Swan neck

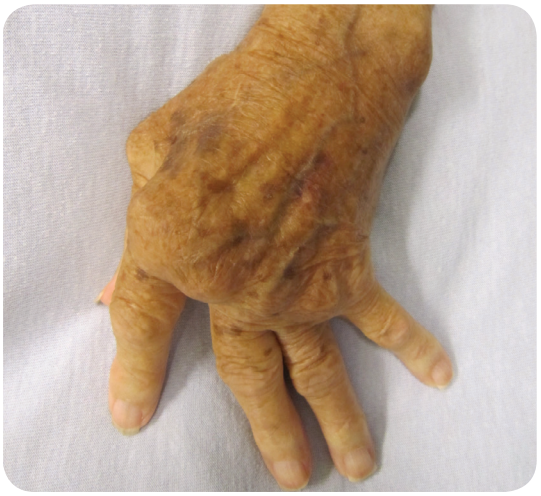

Figure 111.11 Ulnar deviation of the fingers in an individual with rheumatoid arthritis.

Extra-articular manifestations

- Common, systemic signs
  - Fever, fatigue, weight loss
- Rheumatoid nodules
  - Commonly arise on extensor surfaces
- More varied sequelae in severe and/or chronic disease

DIAGNOSIS

DIAGNOSTIC IMAGING

X-ray

- Soft tissue swelling
- Bony erosions
- ↓ Bone density
- Narrowed joint space (late finding)

LAB RESULTS

- RF titers
  - High titers associated with more severe disease
  - Eventually present in 80% of affected individuals
- Anti-citrullinated peptide/protein antibodies (anti-CCP)
  - Sensitivity 50–75%; specificity > 90%
- ↑ ESR, ↑ CRP
- Normocytic anemia (anemia of chronic disease)

OTHER DIAGNOSTICS

History

- Symptoms for > six weeks

Physical examination

- Inflammatory (warm, stiff, painful) arthritis of > three joints
- Characteristic malformations
  - Ulnar deviation, boutonniere ("buttonhole") malformation, swan neck malformation

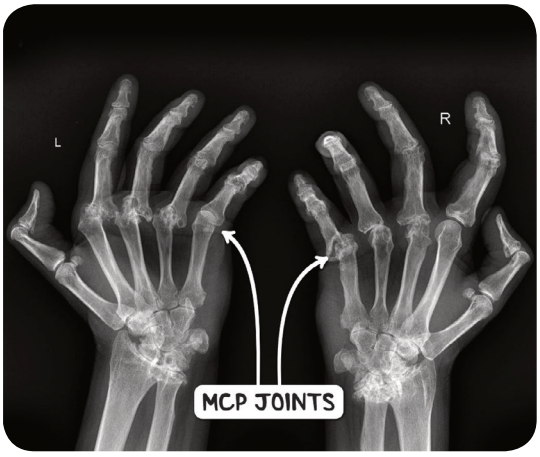

Figure 111.12 An X-ray image of the hands of an individual with rheumatoid arthritis. There is destruction of the metacarpophalangeal joints, the carpometacarpal joints and the wrist.

TREATMENT

MEDICATIONS

- NSAIDs
- Short-term, low-dose glucocorticoid
- DMARD
  - Hallmark of RA treatment
  - Methotrexate (give with folic acid to ↓ side effects)
  - Others: leflunomide, hydroxychloroquine, sulfasalazine
- Biologic DMARDs
  - Adalimumab, etanercept (intercept), infliximab particularly effective (block TNF-alpha, which is thought to underlie most joint damage)

- Abatacept (suppresses T cells)
- Rituximab (suppresses B cells)
- Anakinra (blocks IL-1)
- Tocilizumab (blocks IL-6)

SURGERY

- Only if medication fails
- Severe joint malformation
  - Synovectomy
- Severe malformation, disability
  - Joint replacement

OTHER INTERVENTIONS

- Exercise to maintain range of motion and muscle strength

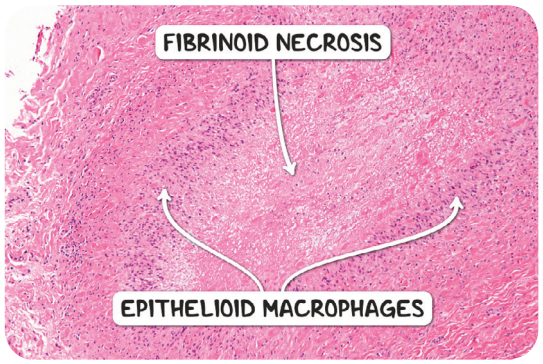

Figure 111.13 A histological section of a rheumatoid nodule. There is granaulomatous inflammation composed of central fibrinoid necrosis and palisading histiocytes.

NOTES
INFLAMMATORY CONNECTIVE TISSUE DISORDERS

GENERALLY, WHAT ARE THEY?

PATHOLOGY & CAUSES

- Chronic autoimmune disorders characterized by inflammation; primarily affect connective tissue
- Production of autoantibodies → deposition of immune complexes → complement activation → tissue destruction
- Inflammatory cytokines stimulate fibroblasts → increased collagen deposition (fibrosis)
- Affects multiple organ systems
  - Skin, heart, respiratory system, urinary, gastrointestinal (GI) tract

CAUSES

- Genetic, environmental factors

COMPLICATIONS

- Skin necrosis; renal, cardiac failure; pulmonary insufficiency; GI reflux/bleeding

SIGNS & SYMPTOMS

- Constitutional symptoms
  - Low grade fever, fatigue, weight loss
- Specific to disease, organ systems affected
  - "Butterfly skin rash" specific to systemic lupus erythematosus (SLE)

DIAGNOSIS

DIAGNOSTIC IMAGING

Barium swallow X-ray

- GI involvement

LAB RESULTS

- Blood tests
  - Hematologic abnormalities, increased inflammatory markers, complications (e.g. increased creatinine reflecting renal failure)
- Serological tests
  - Antibodies, confirm diagnosis

OTHER DIAGNOSTICS

- Physical examination (e.g. characteristic skin rashes)
- Pulmonary function tests
  - Pulmonary involvement

TREATMENT

- Usually symptomatic (e.g. analgesics)

MEDICATIONS

- Steroids/other immunosuppressive agents
  - Reduce inflammation

CREST SYNDROME

osms.it/CREST-syndrome

PATHOLOGY & CAUSES

- Form of limited systemic sclerosis
- Composed of five features; see mnemonic
  - *Calcinosis:* deposition of calcium under skin
  - *Raynaud's syndrome:* episodic, dramatic constriction of arteries in hands
  - *Esophageal dysmotility:* atrophied muscle in esophagus without significant inflammation/fibrosis
  - *Sclerodactyly:* fibrosis of skin of digits
  - *Telangiectasia:* dilation of small blood vessels
- Caused by chronic autoimmune inflammation triggered mainly by anticentromere antibodies (ACAs)
- More benign clinical course than other forms of sclerosis

MNEMONIC: CREST

Features of CREST syndrome

Calcinosis
Raynaud's syndrome
Esophageal dysmotility
Sclerodactyly
Telangiectasia

COMPLICATIONS

- Ischemic ulcers, gangrene, predisposition to chronic skin infections (due to sclerosis, severe ischemia of skin)
- Upper GI bleeding (due to mucosal telangiectasias)

SIGNS & SYMPTOMS

- Calcific nodules under the skin
- White-blue-red transitions in skin color in response to triggers (e.g. low temperature, stress)
- Dysphagia (due to esophageal dysmotility)
- Sclerodactyly
- Telangiectasias (esp. hands, face)

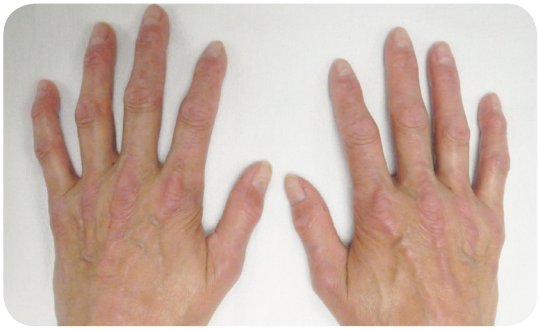

Figure 112.1 Sclerodactyly in an individual with CREST syndrome.

DIAGNOSIS

LAB RESULTS

- Serum blood tests
  - ↑ ANAs: sensitive for systemic sclerosis
  - ↑ ACAs: highly specific (limited systemic sclerosis); confirm diagnosis

OTHER DIAGNOSTICS

- Clinical history, physical examination

TREATMENT

MEDICATIONS

- Steroids
- If sclerosis progresses, stronger immunosuppressants (e.g. cyclosporine)

FIBROMYALGIA

osms.it/fibromyalgia

PATHOLOGY & CAUSES

- Chronic condition of central sensitization; hypersensitivity to pain, sleep disturbances
  - ↓ serotonin (inhibits pain signals)
  - ↑ substance P, ↑ nerve growth factor (involved in propagating pain signals)
  - Predominance in individuals who are biologically female

CAUSES

- Genetic factors
- Environmental factors (child abuse)
- Negative emotions (depression, anxiety, negative beliefs) can amplify pain

SIGNS & SYMPTOMS

- Low threshold to pain
- Widespread muscle pain
- Extreme tenderness in various parts of body
- Sleep disturbances → fatigue, headache
- Difficulty concentrating, remembering things; AKA "fibro fog"

DIAGNOSIS

OTHER DIAGNOSTICS

Diagnostic Criteria

- Pain in ≥ seven areas of body with symptom severity (SS) of ≥ 5 (of 12)/pain in ≥ five areas of body with SS of ≥ 9 (of 12)
- Final score between 0–12
- Symptoms present ≥ three months
- Pain not due to another disorder

Symptom severity (SS) measures

- Fatigue; waking unrefreshed; cognitive symptoms; somatic symptoms
  - *0:* no problem
  - *1:* slight/mild/intermittent
  - *2:* moderate/considerable/often present
  - *3:* severe, continuous, life disturbing

TREATMENT

MEDICATIONS

- If non-pharmacologic measures fail, drug therapy
- Antidepressants
  - Inhibit pain by elevating levels of serotonin, norepinephrine
  - *Tricyclic antidepressants (TCAs):* amitriptyline first line treatment
  - *Serotonin-norepinephrine reuptake inhibitors (SNRIs):* milnacipran
- Anticonvulsants
  - Slow nerve impulses, relieve sleep disturbances

PSYCHOTHERAPY

- Cognitive behavioral therapy (CBT)
  - Manage pain, change negative feelings

OTHER INTERVENTIONS

- Physical therapy, relaxation techniques, sleep hygiene to reduce pain, fatigue

MIXED CONNECTIVE TISSUE DISEASE (MCTD)

osms.it/mixed-connective-tissue-disease

PATHOLOGY & CAUSES

- Overlap autoimmune syndrome; constellation of SLE, systemic sclerosis, polymyositis; may not occur simultaneously
- Can evolve into classic SLE/systemic sclerosis

COMPLICATIONS

- Pulmonary hypertension; interstitial lung disease; renal disease

SIGNS & SYMPTOMS

- Arthralgias (due to polyarthritis)
- Myalgias (due to mild myositis)
- Swollen hands with puffy fingers (due to synovitis)
- Sclerodactyly
- Early development of Raynaud phenomenon
- Fatigue
- Low-grade fevers

DIAGNOSIS

- Confirmation requires characteristic clinical presentation

LAB RESULTS

- High serum levels of anti-U1 ribonucleoprotein (anti-U1-RNP) antibodies
- High ANAs, RF, anti dsDNA, anti Sm, anti Ro

TREATMENT

- Depends on predominant autoimmune disease

MEDICATIONS

- Corticosteroids
  - Suppress immune system

POLYMYALGIA RHEUMATICA (PMR)

osms.it/polymyalgia-rheumatica

PATHOLOGY & CAUSES

- Immune-mediated rheumatic condition affecting joints, sparing muscles
- Most commonly affects shoulder, hip joints
- Usually occurs in individuals who are biologically female > 50; mean age 70
- Strongly associated with giant-cell arteritis, AKA temporal arteritis
- Can regress without treatment after 1–2 years/remain chronic

CAUSES

- *Genetic defects:* specific allele of human leukocyte antigen (HLA)-DR4
- *Environmental factors:* exposure to adenovirus/human parvovirus B19

SIGNS & SYMPTOMS

- Joint pain, stiffness (shoulder, hip joints)
  - Often starts unilaterally, progresses to bilateral within few weeks
  - More severe after prolonged inactivity (e.g. morning)
  - Typically lasts > one hour
  - Affects nearby nerves in muscle → muscle pain (referred pain)
- Constitutional symptoms
  - Low grade fever (interleukins act as pyrogens)
  - Fatigue
  - Loss of appetite → weight loss
- If severe headache, jaw pain, vision problems
  - Temporal arteritis

DIAGNOSIS

LAB RESULTS

- Increased serum inflammatory markers
  - Erythrocyte sedimentation rate (ESR)
  - C-reactive protein (CRP)
- Biopsy
  - Inflammation in joints

OTHER DIAGNOSTICS

- Physical examination
  - Decreased passive range of motion of affected joints

TREATMENT

MEDICATIONS

- Low dose of corticosteroids
  - Suppress immune response

RAYNAUD'S DISEASE

osms.it/raynauds-disease

PATHOLOGY & CAUSES

- Vasospasm of skin arteries in response to triggers, resulting in skin color transitions
- Exposure to trigger → stimulation of sympathetic nerves in arteriole walls → vasospasm of arterioles → decrease in blood flow
- Usually affects hands, fingers, toes; can affect nose, ears, lips
- Common triggers
  - Emotional stress; low temperatures; nicotine; caffeine; medications that affect sympathetic nervous system (e.g. pseudoephedrine)

TYPES

Primary: Raynaud phenomenon/disease

- Common in pregnant individuals, people who work in jobs involving vibration (e.g. jackhammer)

Secondary: Raynaud syndrome

- Connective tissue disorders
  - Systemic lupus erythematosus (SLE), scleroderma, mixed connective tissue disease
- Disorders affecting blood vessels
  - Buerger's disease, Takayasu's arteritis, thromboangiitis obliterans
- Medications
  - Beta blockers, nicotine

COMPLICATIONS

- Ulceration, infarction, tissue necrosis, gangrene (if severe)

SIGNS & SYMPTOMS

- Vasospasm → changes in skin color of hands, fingers, toes
  - *White:* ischemia
  - *Blue:* hypoxia after prolonged ischemia
  - *Red:* reactive hyperemia (vasospasm ends, oxygenated blood rushes into tissue)
- Raynaud phenomenon
  - Affects hand fingers, toes symmetrically; severity remains constant
- Raynaud syndrome
  - Asymmetrical; progressive severity
- Swelling, numbness, tingling, pain (due to reactive hyperemia)

DIAGNOSIS

- Based upon description of episodes

DIAGNOSTIC IMAGING

- Nailfold capillary microscopy to examine finger capillaries
  - *Normal appearance:* Raynaud phenomenon
  - *Damaged appearance:* Raynaud syndrome

TREATMENT

MEDICATIONS

- Vasodilators (e.g. calcium channel blockers)

SURGERY

- If severe, surgery to cut sympathetic nerve fibers supplying affected areas

OTHER INTERVENTIONS

- Avoid triggers

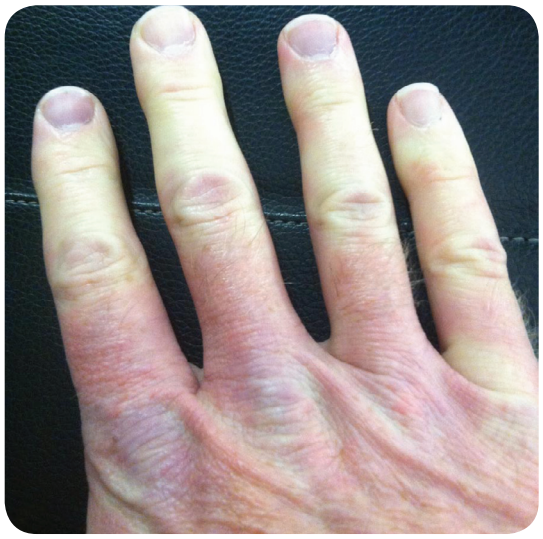

Figure 112.2 A hand with pale fingers caused by Raynaud's disease.

SCLERODERMA

osms.it/scleroderma

PATHOLOGY & CAUSES

- AKA systemic sclerosis
- Chronic inflammatory autoimmune disease, can result in widespread damage to small blood vessels, excessive fibrosis
  - T helper cells activated by unknown antigen → release cytokines → stimulate inflammatory cells, fibroblasts → chronic inflammation, excessive collagen deposition
  - Mediators released by inflammatory cells → damage microvasculature → ischemic injuries, scarring
- Primarily affects skin, can involve visceral organs
  - GI tract, kidneys, heart, muscles, lungs

TYPES

Limited (80%)

- Skin involvement limited to fingers, forearms, face
- Late visceral involvement
- Some individuals develop CREST syndrome
  - Calcinosis, Raynaud's phenomenon, esophageal dysmotility, sclerodactyly, telangiectasia
- Associated with anticentromere antibodies
- Relatively benign

Diffuse (20%)

- Widespread skin involvement
- Early visceral involvement
- Rapid progression
- Associated with anti-DNA topoisomerase I antibodies
- Poor prognosis

RISK FACTORS

- More common in individuals who are biologically female (3:1 ratio)
- Average age of onset: 35–50
- Genetic factors
- Environmental factors (e.g. viruses, toxins, drugs)

COMPLICATIONS

- Excessive skin fibrosis → painful ulcers, disfigurement, disability
- Severe internal organ involvement → renal, cardiac failure; pulmonary insufficiency; intestinal malabsorption

SIGNS & SYMPTOMS

- Raynaud phenomenon
  - Precedes other symptoms, present in almost all individuals
- Cutaneous changes of face, extremities
  - Skin thickening, tightening, sclerosis (most common); edema, erythema (precede sclerosis)
- GI involvement
  - Esophageal fibrosis → dysphagia, GI reflux
  - Small intestine involvement → abdominal pain, obstructions, constipation, diarrhea, malabsorption syndrome (weight loss, anemia)
- Pulmonary involvement with interstitial fibrosis
  - Right-sided cardiac dysfunction/ pulmonary hypertension
- Cardiac involvement
  - Pericardial effusions, myocardial fibrosis → congestive heart failure, arrhythmias
- Renal involvement (diffuse disease) → fatal hypertensive crisis (rare)

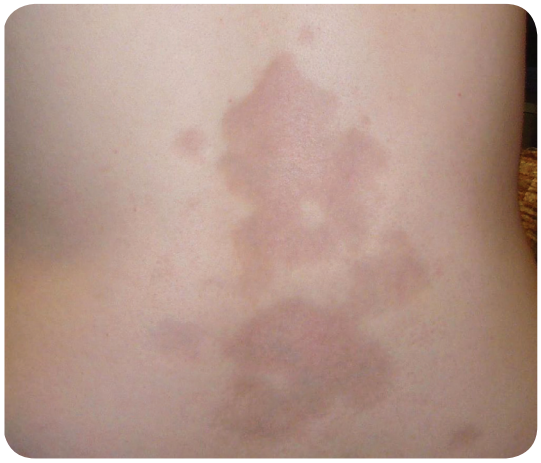

Figure 112.4 A rash on the back of an individual with a form of localised scleroderma known as morphea.

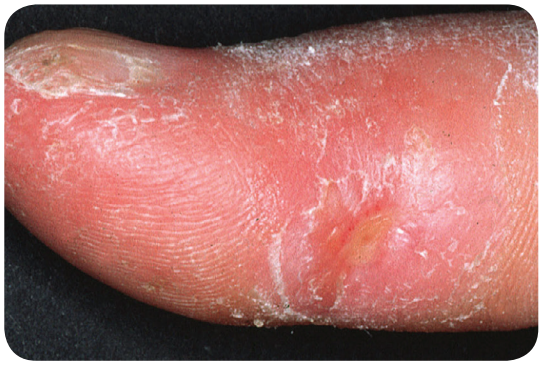

Figure 112.3 The finger of an individual with systemic sclerosis showing sclerosis, erythema and ulcer formation.

DIAGNOSIS

DIAGNOSTIC IMAGING

- Upper endoscopy
  - Esophageal fibrosis/reflux esophagitis

LAB RESULTS

- Serologic tests
  - ↑ ANAs in almost all individuals with systemic sclerosis; low specificity
  - ↑ ACAs highly specific (limited)
  - Anti-topoisomerase I antibodies (anti-Scl-70) highly specific (diffuse)
- Complete blood count (CBC)
  - Anemia due to malabsorption, increased serum creatinine due to renal dysfunction

OTHER DIAGNOSTICS

- Clinical presentation
  - Skin thickening, swollen fingers, Raynaud's phenomenon, GI reflux
- Pulmonary function tests
  - Restrictive ventilatory defect due to pulmonary interstitial fibrosis

TREATMENT

- Depends on disease subset, severity of internal organ involvement

MEDICATIONS

- Usually symptomatic
  - Analgesics for musculoskeletal pain
  - Proton pump inhibitors for gastroesophageal reflux
  - Calcium channel blockers for Raynaud's phenomenon
  - Angiotensin converting enzyme (ACE) inhibitors for renal hypertensive crisis
- *Immunosuppressive therapy initiation:* diffuse skin/severe internal organ involvement

SJOGREN'S SYNDROME (SS)

osms.it/sjogrens-syndrome

PATHOLOGY & CAUSES

- Chronic autoimmune inflammatory disease; lymphocytic infiltration, destruction of exocrine glands of eyes, mouth
- Proposed mechanisms
  - Immune reactions against antigens of viral infection of exocrine glands
  - Autoimmune T cell reaction against unknown self antigen expressed in salivary, lacrimal glands
- Variety of extraglandular manifestations may occur
- Usually occurs in individuals who are biologically female, 50–60 years

CAUSES

- *Primary:* sicca syndrome
- *Secondary* (to other autoimmune diseases)*:* rheumatoid arthritis (most common)

COMPLICATIONS

- Periodontal complications; oral infections; mucosal associated lymphoid tissue (MALT) lymphoma

SIGNS & SYMPTOMS

- Dry eyes
  - Irritation, itching, foreign body sensation, keratoconjunctivitis
- Oral dryness reflecting salivary hypofunction
- Salivary gland enlargement (parotid, submandibular, etc.)
- Extraglandular manifestations
  - Musculoskeletal symptoms (arthralgias, arthritis); rashes; interstitial nephritis, vasculitis

DIAGNOSIS

- *Clinical presentation:* persistent dry eyes/mouth, parotid gland enlargement

DIAGNOSTIC IMAGING

Parotid gland MRI

- Honeycomb pattern

Salivary gland ultrasound

- Multiple hypoechoic areas

LAB RESULTS

- CBC
  - Leukopenia, thrombocytopenia, anemia
- ↑ ESR
- Urinalysis

- Proteinuria/hematuria reflecting glomerulonephritis
- Labial salivary gland biopsy (confirm diagnosis)
  - Focal lymphocyte foci (collections of tightly aggregated lymphocytes)
- Serologic tests (support diagnosis)
  - ↑ antinuclear antibodies (ANAs) in 95% of individuals
  - ↑ rheumatoid factor (RF) in 50–75% of individuals with/without rheumatoid arthritis
  - Anti-Sjögren syndrome A (SSA) (Ro), Anti-Sjögren syndrome B (SSB) (La) specific to SS, found elevated only in 55%, 40% of individuals, respectively

OTHER DIAGNOSTICS

Tear deficiency tests

- Schirmer test
  - Measures reflex tear production; wetting of test paper < 5mm indicative of tear deficiency
  - Ocular surface staining with Rose Bengal stain and slit-lamp examination—assess tear break-up time (TBUT); TBUT < 10 seconds indicative of tear deficiency
- Salivary gland tests
  - *Salivary gland scintigraphy:* low uptake of radionuclide characteristic of SS
  - *Sialometry:* low volume of saliva indicative of salivary gland hypofunction

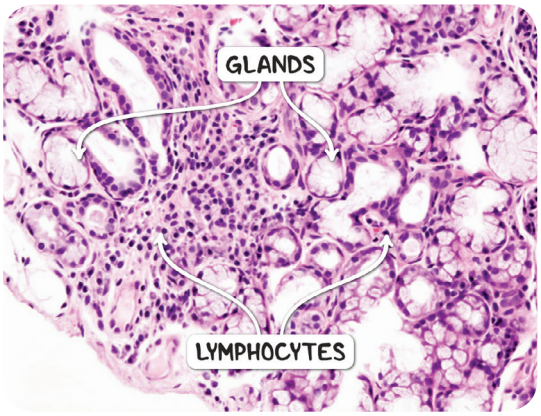

Figure 112.5 A lymphocytic infiltrate in a minor salivary gland excised from an individual with Sjögren's syndrome.

TREATMENT

MEDICATIONS

- Mild SS
  - Secretagogues
  - Local treatment for ocular, oral dryness (e.g. artificial tears)
- Moderate to severe SS
  - Immunosuppressive treatment

SYSTEMIC LUPUS ERYTHEMATOSUS (SLE)

osms.it/systemic-lupus-erythematosus

PATHOLOGY & CAUSES

- Chronic systemic autoimmune disorder; wide range of clinical, serological features
- Periods of flare-ups, remittance
- Environmental triggers damage DNA → apoptosis → release of nuclear bodies
- Clearance of apoptotic bodies ineffective due to genetic defects → increased amount of nuclear antigens in bloodstream → initiates immune response → production of antinuclear antibodies → bind to antigens, form immune complexes
- Complexes deposit in tissues (e.g. kidneys, skin, joints, heart) → Type III hypersensitivity reaction
- Individuals may develop antibodies targeting molecules (e.g., phospholipids) of red, white blood cells → marking them for phagocytosis → Type II hypersensitivity reaction

RISK FACTORS

- Genetic defects associated with SLE
- UV radiation
- Smoking
- Viral, bacterial infections
- Medications (e.g. procainamide, hydralazine, isoniazid, estrogens)
- More common in individuals who are biologically female, of reproductive age

COMPLICATIONS

- Cardiovascular disease
  - Libman–Sacks endocarditis, myocardial infarction (MI)
- Serious infections; renal failure; hypertension
- Antiphospholipid syndrome
  - Hypercoagulable state; individuals prone to develop clots (e.g. deep vein thrombosis, hepatic vein thrombosis, stroke)

SIGNS & SYMPTOMS

- Fever, joint pain, rash in sun-exposed areas
- Typical rashes
  - *Malar rash (butterfly rash):* over cheeks
  - *Discoid rash:* plaque-like/patchy redness, can scar
  - *General photosensitivity:* typically lasts few days

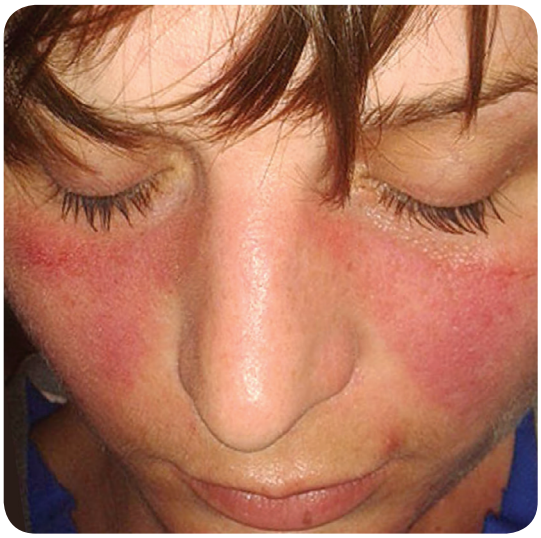

Figure 112.6 A butterfly rash on the face of an individual with systemic lupus erythematosus.

- Weight loss
- Ulcers in oral/nasal mucosa
- Serositis (e.g. pleuritis/pericarditis)
- Libman–Sacks endocarditis: formation of nonbacterial vegetations on ventricular, atrial valve surfaces; mitral, aortic valves (most common)
- Myocarditis
- Renal disorders
  - Abnormal levels of urine protein, diffuse proliferative glomerulonephritis
- Neurologic disorders
  - Seizures, psychosis
- Hematologic disorders
  - Anemia, thrombocytopenia, leukopenia

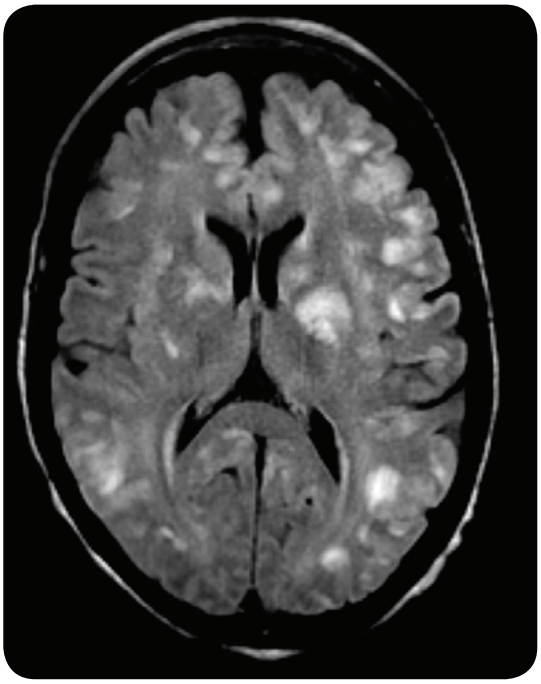

Figure 112.7 An MRI scan of the head of an individual with SLE who presented with altered mental status and seizures. There a numerous small infarcts suggestive of cerebral vasculitis. The individual improved after treatment with steroids.

DIAGNOSIS

OTHER DIAGNOSTICS

Diagnostic criteria (4 of 11)

- Malar rash
- Discoid rash
- General photosensitivity
- Oral/nasal ulcers
- Serositis
- Arthritis in ≥ two joints
- Renal disorders
- Neurologic disorders
- Hematologic disorders
- Antinuclear antibodies
  - Very sensitive, not specific
- Other antibodies
  - *SLE specific:* anti-Smith, anti-dsDNA
  - *Anti-phospholipid:* anticardiolipin (false-positive test for syphilis); lupus anticoagulant (lupus antibody); anti-beta 2 glycoprotein I

TREATMENT

- *Goal:* prevent relapses, limit severity

MEDICATIONS

- Long term therapy
  - Antimalarial agents
- Mild to moderate manifestations
  - Non-steroidal anti-inflammatory drugs (NSAIDs), low doses of corticosteroids
- Severe/life-threatening manifestations
  - High doses of corticosteroids, intensive immunosuppressive drugs

OTHER INTERVENTIONS

- Avoid sun exposure
- Physical exercise
- Balanced diet
- Smoking cessation
- Immunizations

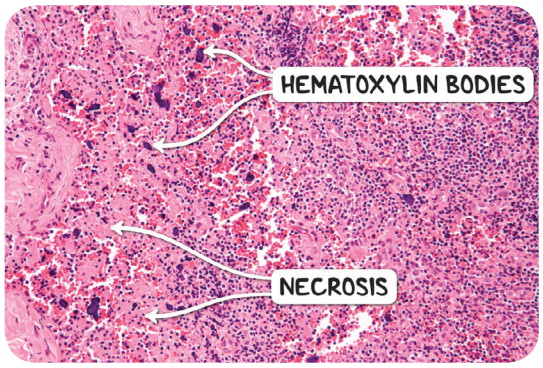

Figure 112.8 A histological section of a lymph node from an individual with lupus lymphadenopathy. There is necrosis, with an absence of neutrophils, and large numbers of hematoxylin bodies.

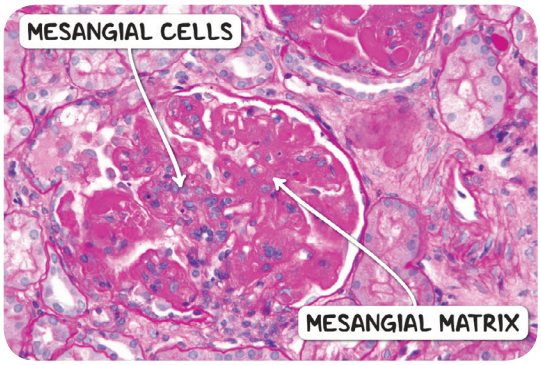

Figure 112.9 Histological appearance of the glomerulus in a case of lupus nephritis. There is global mesangial cell proliferation and abundant mesangial matrix.

NOTES
INFLAMMATORY MYOSITIS

GENERALLY, WHAT IS IT?

PATHOLOGY & CAUSES

- Multiple disorders involving autoimmune inflammation, injury of skeletal muscles
- Most commonly include polymyositis, dermatomyositis, inclusion body myositis
  - Dermatomyositis predominantly mediated by humoral immune response; polymyositis, inclusion body myositis by cellular immune response

RISK FACTORS

- Age (more common in older population)
- Dermatomyositis, polymyositis more common in individuals who are biologically female
- Inclusion body myositis more common in individuals who are biologically male
- Chronic viral infections: human T cell lymphotropic virus Type I (HTLV-1), HIV
- Autoimmune diseases
- Malignancies

COMPLICATIONS

- Dysphagia, pulmonary involvement
- Cardiovascular involvement

SIGNS & SYMPTOMS

- Proximal muscle weakness
- Inclusion body myositis
  - Distal muscle weakness
- Dermatomyositis
  - Skin rashes

DIAGNOSIS

LAB RESULTS

- ↑ muscle enzymes, like creatine kinase (CK)
- Muscle biopsy
  - *Dermatomyositis:* perivascular, perimysial inflammation
  - *Polymyositis:* endomysial inflammation
  - *Inclusion body myositis:* endomysial inflammation, intracytoplasmic vacuoles with protein depositions

OTHER DIAGNOSTICS

- Physical examination
  - Muscle weakness

Electromyography (EMG)

- Pathological signals

TREATMENT

MEDICATIONS

- Corticosteroids, immunosuppressive agents

OTHER INTERVENTIONS

- Physical rehabilitation

DERMATOMYOSITIS

osms.it/dermatomyositis

PATHOLOGY & CAUSES

- Autoimmune disorder leading to destruction of small blood vessels in muscles, skin
- Unknown factor activates C3 protein (complement component 3) → formation of membrane attack complex (MAC), accumulation in capillaries → destruction of capillary wall → microinfarctions
- *Juvenile:* around seven years; associated with calcinosis (deposition of calcium in skin)
- *Adult:* > 40; associated with malignancy, treating malignancy may cure myositis

RISK FACTORS

- > 60 years
- Malignancy

COMPLICATIONS

- Respiratory muscle weakness; dysphagia (if esophagus, pharyngeal muscles involved); interstitial pulmonary disease; cardiovascular involvement

SIGNS & SYMPTOMS

- Weakness starts in proximal muscles, slowly progresses (e.g. difficulty getting up)
- Heliotrope rash
  - Purplish eyelids with posible periorbital edema
- Gottron papules
  - Scaling erythema of knuckles, elbow, knees
- V-shaped rash on chest

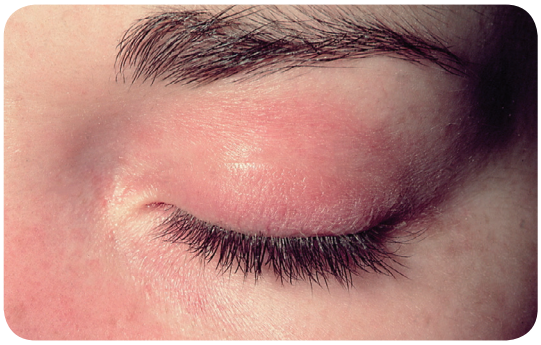

Figure 113.1 A heliotrope rash affecting the eyes of an individual with dermatomyositis.

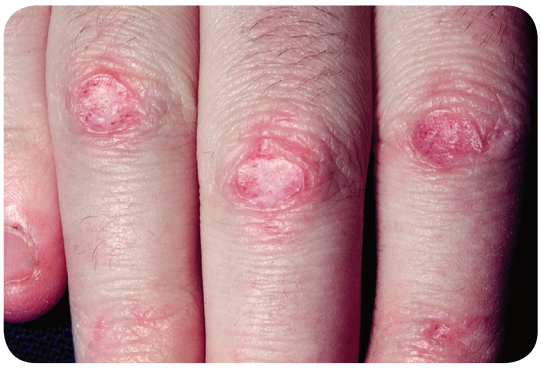

Figure 113.2 Gottron's papules on the extensor surfaces of an individual with dermatomyositis.

DIAGNOSIS

DIAGNOSTIC IMAGING

CT scan

- Malignancy suspected

LAB RESULTS

- Blood tests
  - ↑ CK (muscle cells death)
  - ↑ aspartate aminotransferase (AST)

- ↑ lactic dehydrogenase (LDH)
- Antinuclear antibodies (ANA)
- Anti-Mi-2 antibodies (acute phase, better prognosis)

- Biopsy
  - Perivascular, perimysial inflammation
  - Perifascicular atrophy
  - "Ghost fibers" (destroyed fibers, can no longer be stained)

OTHER DIAGNOSTICS

EMG

- Abnormal signals

TREATMENT

MEDICATIONS

- Corticosteroids (e.g. glucocorticoid)
- Immunosuppressive agents (e.g. methotrexate)
- IV immune globulins

OTHER INTERVENTIONS

- Physical therapy (preserve muscle strength)
- Sunscreen, avoid sun exposure (in skin disease)

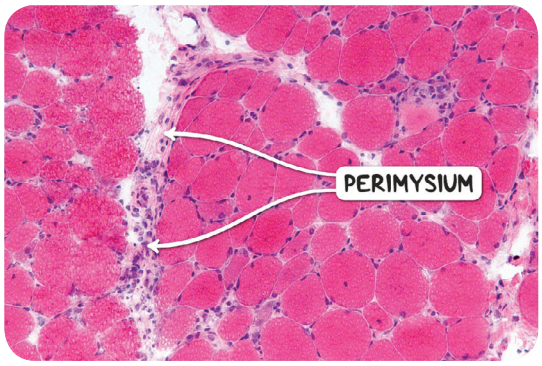

Figure 113.3 The histological appearance of the skeletal muscle of an individual with dermatomyositis. The perimysium and endomysium have been infiltrated by chronic inflammatory cells, with predilection for the perimysium.

INCLUSION BODY MYOSITIS

osms.it/inclusion-body-myositis

PATHOLOGY & CAUSES

- Idiopathic inflammation of muscles leading to weakness, muscle atrophy
- Inflammation, degenerative processes
- Unknown factor causes myofibers to present major histocompatibility complex class I (MHC I) → CD8+ T cells gather, recognize MHC I, bind → express perforin → pores form on myofibers membranes → cell degeneration
- Accumulation of abnormal amyloidogenic proteins (e.g. beta-amyloid), cytotoxic effect
  - *Causes:* misfolding of proteins; damaged/inhibited proteasomes; endoplasmic reticulum stress

RISK FACTORS

- Age > 50
- *Chronic viral infections:* HTLV-1
- *Autoimmune diseases:* Sjögren's syndrome

COMPLICATIONS

- Dysphagia (if esophagus, pharyngeal muscles involved)

SIGNS & SYMPTOMS

- Slowly progressive muscle weakness, sometimes asymmetric
  - Proximal leg muscles (difficulty getting up, frequent falls)
  - Distal arm muscles (weak grip)

- As disease progresses
  - ↑ muscle atrophy
  - ↓ deep tendon reflexes

DIAGNOSIS

LAB RESULTS

- Mild ↑ muscle enzymes (e.g. CK)
- Muscle biopsy
  - CD8+ T lymphocytes, macrophages infiltrating non-necrotic myofibers
  - Vacuoles with amyloides, other protein accumulations (inclusion bodies)
  - ↑ MHC I on immunostaining

OTHER DIAGNOSTICS

- Clinical presentation
  - Muscle weakness

EMG

- Polyphasic motor unit action potentials (MUAPs) with small amplitude, short duration

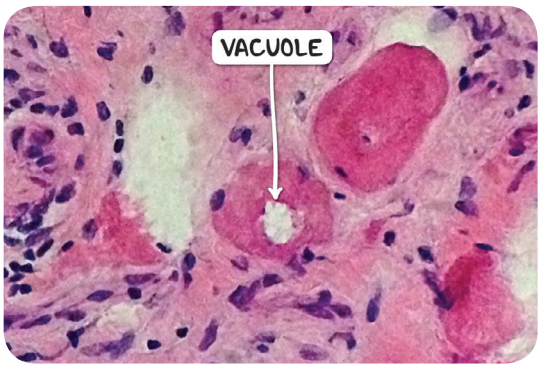

Figure 113.5 A histological section of muscle showing a myofiber vacuole in an individual with inclusion body myositis.

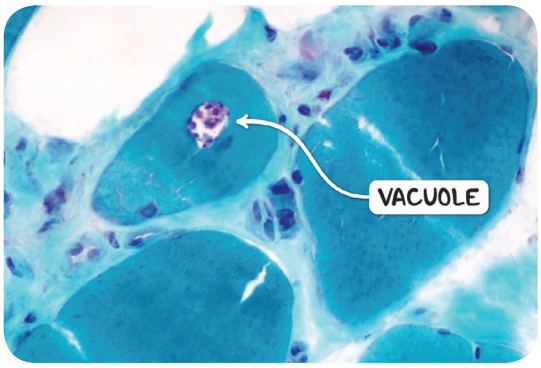

Figure 113.4 Gomori staining highlights the rimmed vacuoles in inclusion body myositis.

TREATMENT

MEDICATIONS

- Immunosuppressive therapy
  - Administered when another systemic autoimmune disease present

OTHER INTERVENTIONS

- Physical therapy
  - Muscle strengthening
- Speech therapy
  - If dysphagia present
- Occupational therapy

POLYMYOSITIS

osms.it/polymyositis

PATHOLOGY & CAUSES

- Inflammatory destruction of muscles leading to muscle weakness
- Unknown factor induces CD8+ T cells, macrophages to recognize nuclear, cytoplasmic antigens of muscle cells → immune cells surround nonnecrotic muscle cells → muscle cell destruction

RISK FACTORS

- Autoimmune disease
- Chronic viral infection (HIV, HTLV-1)

COMPLICATIONS

- Aspiration pneumonia
- Interstitial lung disease
- Dysphagia → malnutrition, anorexia

SIGNS & SYMPTOMS

- Symmetrical weakness of proximal leg, arm muscles (e.g. difficulty climbing stairs)
- Neck flexor weakness
- Mild myalgia, tenderness
- Dysphagia (if esophagus, pharyngeal muscles involved)

DIAGNOSIS

DIAGNOSTIC IMAGING

Chest X-ray, CT scan

- Pulmonary involvement

LAB RESULTS

- Blood tests
  - ↑ CK, aldolase; ANA; antisynthetase antibodies (anti-Jo-1)
- Muscle biopsy
  - Endomysial inflammation; intact blood vessels; myofibers surrounded by CD8+ T lymphocytes, macrophages

OTHER DIAGNOSTICS

- Physical examination
  - Muscle weakness, tenderness

EMG

- Low amplitude, short duration potential; repetitive discharges

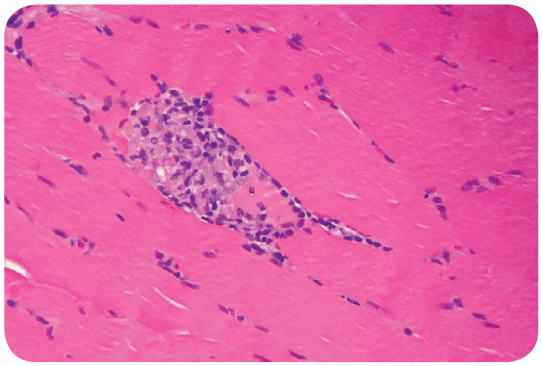

Figure 113.6 A muscle biopsy from an individual with polymyositis. The lymphocytes penetrate individual myofibers. In this example, the inflammation has progressed to phagocytic destruction by macrophages.

TREATMENT

MEDICATIONS

- Corticosteroids
- Immunosuppressive agents (if nonresponsive to corticosteroids)
- IV immune globulins (if severe, life-threatening)

OTHER INTERVENTIONS

- Physical therapy (preserve muscle strength)

INFLAMMATORY MYOSITIS SUMMARY

	DERMATOMYOSITIS	INCLUSION BODY MYOSITIS	POLYMYOSITIS
MAIN IMMUNE RESPONSE	Humoral	Cellular	Cellular
HISTOLOGIC FINDINGS	Perimysial, perivascular inflammation	Endomysial inflammation, inclusion bodies	Endomysial inflammation
SEX PREDISPOSITION	Individuals who are biologically female	Individuals who are biologically male	Individuals who are biologically female
MANIFESTATION	Proximal muscle weakness, skin rash	Proximal, distal muscle weakness	Proximal muscle weakness
MUSCLE ENZYMES	↑ CK	Slightly ↑ / normal CK	↑ CK
TREATMENT	Corticosteroids, immunosuppressive agents	Nonresponsive	Corticosteroids, immunosuppressive agents

NOTES
JOINT PATHOLOGY

GENERALLY, WHAT IS IT?

PATHOLOGY & CAUSES

- Disorders affecting joints
- Most commonly caused by trauma

SIGNS & SYMPTOMS

- Asymptomatic or pain during rest/movement

DIAGNOSIS

DIAGNOSTIC IMAGING
- Radiography
- MRI

LAB RESULTS
- Synovial fluid analysis

TREATMENT

- Treat symptoms pharmacologically
- Surgical procedures

BAKER'S CYST

osms.it/bakers-cyst

PATHOLOGY & CAUSES

- Synovial fluid accumulates in popliteal bursa (between medial head of gastrocnemius, semimembranosus muscles) → swelling
- *Adults*: popliteal bursa communicates with synovial sac; underlying knee joint disease main cause
  - Knee joint disease → ↑ synovial fluid production → synovial fluid squeezes through valve-like formation into bursa → fluid unable to flow backward → bursa enlarges → lump-like structure in the popliteal fossa
- *Children*: noncommunicating cyst; usually arises as primary process

CAUSES
- Chronic knee joint trauma
- Osteoarthritis
- Rheumatoid arthritis
- Meniscal tears

COMPLICATIONS
- Cyst enlargement
  - In popliteal space → obstruction of veins → lower leg swelling
  - Extension to calf → swelling, redness, bruising, positive Homan's sign (calf pain during dorsiflexion of the foot) → similar to deep-vein blood clot
- Rupture

SIGNS & SYMPTOMS

- May be asymptomatic
- Stiffness and pain in the knee → worse with prolonged standing

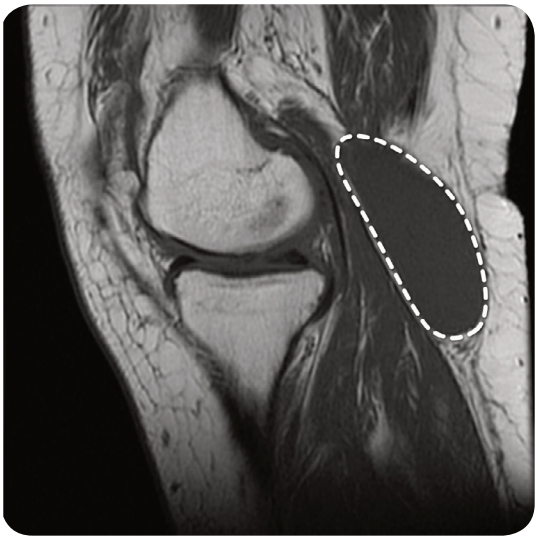

Figure 114.1 An MRI scan of the knee joint in the sagittal plane demonstrating a Baker's cyst in the popliteal fossa.

DIAGNOSIS

DIAGNOSTIC IMAGING

Ultrasound and MRI

- Fluid-filled cyst; differentiation between cyst, blood clot

X-ray

- Bone, joint pathology associated with cyst

OTHER DIAGNOSTICS

- Physical examination
  - Lump in the back of the knee

TREATMENT

SURGERY

- Surgical excision

OTHER INTERVENTIONS

- Fluid aspiration, glucocorticoid intra-articular injection → ↓ size and inflammation
- Treat complications
  - Leg elevation, resting, analgesics

BURSITIS

osms.it/bursitis

PATHOLOGY & CAUSES

- Inflammation of bursa (small sac located between muscles, tendons, bone structures)
- Inflammation of bursa → ↑ production of synovial fluid → enlargement of bursa → ↑ friction during movement → symptomatology
- Most commonly affected bursas
  - Subacromial, olecranon, trochanteric, prepatellar, infrapatellar

CAUSES

- Autoimmune disorders
  - Rheumatoid arthritis, ankylosing spondylitis, scleroderma, systemic lupus erythematosus → chronic course
- Overuse/trauma, gout, bacterial infections (septic bursitis) → acute course

SIGNS & SYMPTOMS

- Joint pain; stiffness of joints; surrounding skin red
- Acute bursitis
  - Tenderness, pain during activation of muscles adjacent to inflamed bursa
- Chronic bursitis
  - Swelling with minimal pain

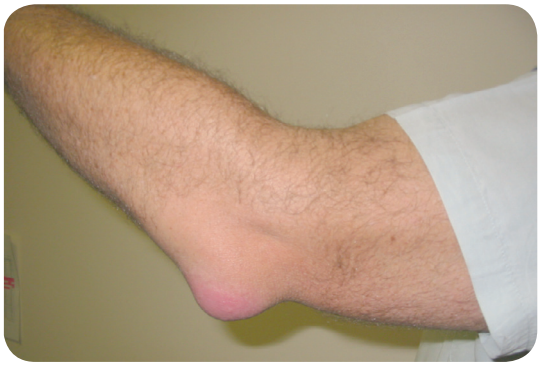

Figure 114.2 An individual with olecranon bursitis.

DIAGNOSIS

DIAGNOSTIC IMAGING

Ultrasound

- Differentiation from Baker's cyst

LAB RESULTS

- Aspiration and analysis of synovial fluid
  - *Infection*: ↑ polymorphonuclear leukocytes, proteins, ↓ glucose
  - *Gout*: ↑ monosodium urate crystals

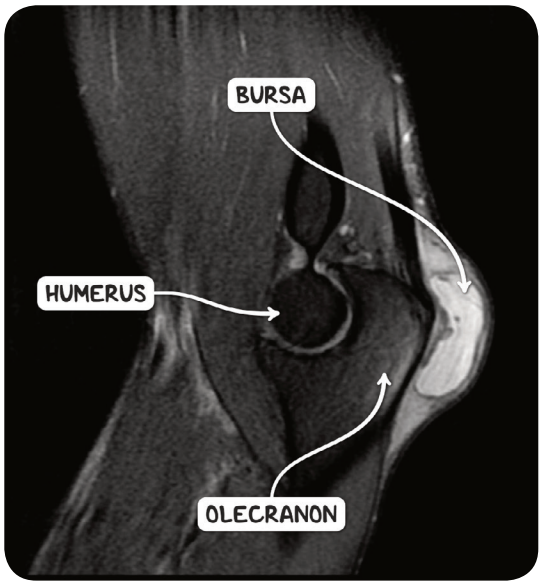

Figure 114.3 An MRI scan of the elbow demonstrating a high signal fluid collection in the olecranon bursa in an individual with olecranon bursitis.

TREATMENT

MEDICATION

- Non-steroidal inflammatory drugs (NSAIDs)
- Injection of steroids, local anesthetics
- Septic bursitis
  - Antibiotics

SURGERY

- Surgical excision
  - Chronic or recurrent bursitis

OTHER INTERVENTIONS

- Resting, elevation

OSTEOARTHRITIS

osms.it/osteoarthritis

PATHOLOGY & CAUSES

- Progressive loss of articular cartilage, underlying bone of synovial joints
- Articular cartilage damage → chondrocytes replace type II collagen with type I, ↓ proteoglycans → eventual exhaustion, apoptosis of chondrocytes → ↓ elasticity, ↑ cartilage breakdown → clefts in articular surface (fibrillations), "joint mice" in synovial space with inflammation of synovium → bone exposition → rubbing other bone → eburnation (polished ivory look)
- Due to damage/inflammation, new bone formation on edges of bone with outward growth → osteophyte (enlargement of the joint with a knob-like look)
  - *Bouchard nodes*: proximal interphalangeal finger joints affected
  - *Heberden nodes*: distal interphalangeal finger joints affected
- Most commonly affected joints
  - Lower spine, hip, knee, foot and hand joints

CLASSIFICATION

- Primary
  - Usually idiopathic
- Secondary
  - Caused by some other condition (e.g. diabetes, alkaptonuria, hemochromatosis, chronic joint injury)

RISK FACTORS

- Aging
  - Cartilage thinning with ↓ hydratation → protein accumulation, collagen crosslinking → cartilage is more breakable; ↑ calcification of meniscus, cartilage
- Inflammation → ↑ proinflammatory cytokines
  - IL1, IL6, TNF → ↑catabolism/↓ anabolism of cartilage
- Obesity
  - Excessive load, metabolic disorders affect joints
- Genetic disorders
  - Mutations in cartilage building collagens (types II, IX and XI)
- Biological sex
  - Biologically female more prone
- Previous joint injuries
- Infection
- Neurologic disorders

COMPLICATIONS

- Cystic degeneration of subchondral bone
- Surrounding ligaments, neuromuscular abnormalities

SIGNS & SYMPTOMS

- Sharp pain/burning sensation worsened by prolonged activity
- Limited range of motion
- Morning stiffness > one hour
- No swelling

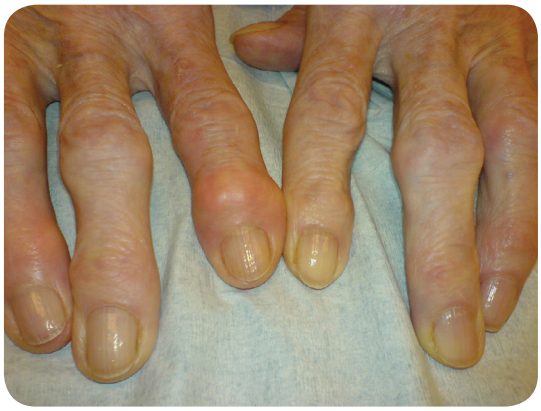

Figure 114.4 Heberden's node on the distal interphalangeal joint of the right index finger in an individual with osteoarthritis.

DIAGNOSIS

DIAGNOSTIC IMAGING

Radiography

- Loss of joint space
- Subchondral bone sclerosis

MRI

- Loss of joint space
- Subchondral bone sclerosis
- Osteophytes
- Visualisation of articular cartilage, surrounding soft tissues

CT scan

- Displacement of foot, ankle, patellofemoral joint

Bone scan

- Detect abnormalities

LAB RESULTS

- Arthrocentesis

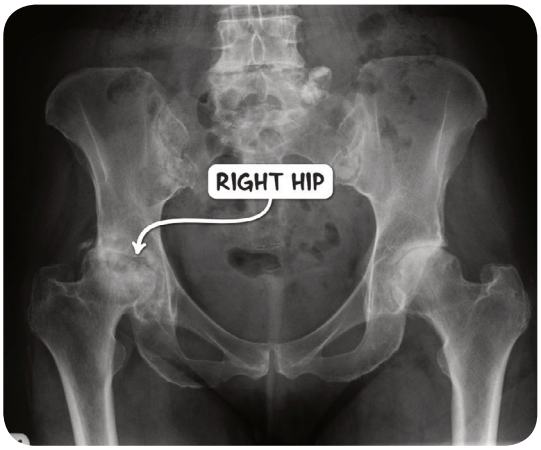

Figure 114.5 An X-ray image of the pelvis demonstrating osteoarthritis of the right hip joint. The femoral head is malformed, there is marked loss of joint space and there are numerous subchondral bone cysts.

TREATMENT

MEDICATIONS

- Pain management
  - Acetaminophen, tramadol, topical and oral non-steroidal anti-inflammatory drugs (NSAIDs)
- Intra-articular injections
  - Corticosteroids
  - Sodium hyaluronate

SURGERY

- Osteotomy
  - Individuals < 60 years with malalignment of hip, knee joint
- Arthroplasty
- Stem-cell therapy

OTHER INTERVENTIONS

- Exercise
- Weight loss
- Physical therapy
- Electromagnetic field stimulation for individuals with knee osteoarthritis

SLIPPED CAPITAL FEMORAL EPIPHYSIS

osms.it/slipped-capital-femoral-epiphysis

PATHOLOGY & CAUSES

- Anterior displacement of femoral head metaphysis, with epiphysis remaining in hip acetabulum
- Caused by growth plate (physis) fracture
- Example of type I Salter–Harris fracture usually affecting one hip
- Hypertrophy of growth plate → abnormal endochondral ossification, cartilage maturation → growth plate weakness → if too much force generated across growth plate → slippage

CLASSIFICATION

- Based on disease course
  - *Acute*: > three weeks
  - *Chronic*: < three weeks
  - *Acute on chronic*: chronic with acute exacerbations
- Based on lesion stability
  - *Stable*: walking possible with/without crutches
  - *Unstable*: walking impossible, even with crutches
- Displacement of the femoral head from neck; seen on radiography
  - *Type I*: slippage < 33%
  - *Type II*: 33–50%
  - *Type III*: > 50%

RISK FACTORS

- Obesity
- ↓ thyroid, growth hormone
- Osteodystrophy
- Down syndrome
- Demographics
  - Adolescent black males of African descent most commonly affected

COMPLICATIONS

- Osteoarthritis
- Metaphysis slippage → ↓ blood flow → avascular necrosis
- Secondary SCFP affecting other hip; usually within a year of first SCFP
- *Unstable displacement*: ↑ complication rate

SIGNS & SYMPTOMS

- Hip, groin, knee pain
- Duck-like gait
- Hip in external rotation, flexion

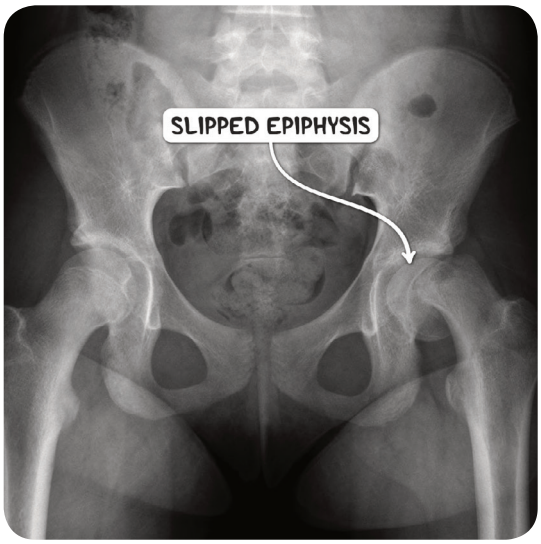

Figure 114.6 An X-ray image of the pelvis demonstrating a slipped capital femoral epiphysis on the left side.

DIAGNOSIS

DIAGNOSTIC IMAGING

X-ray

- Anteroposterior X-ray
  - Melting ice cream cone appearance visible through line of Klein (virtual line parallel to femoral neck's upper edge)
- Frog-leg X-ray
  - Straight line through center of femoral neck anterior to epiphysis (rather than central)

MRI, CT scan

- Accurate measurements of displacement degree

TREATMENT

SURGERY

- Fixation with a cannulated screw
- Preventive fixation of the other hip
  - Children with SCFP before the age of 10
  - Persons with endocrinopathies
- Osteotomy

TRANSIENT SYNOVITIS

osms.it/transient-synovitis

PATHOLOGY & CAUSES

- Inflammation of hip joint synovial membrane
- Cause relatively unknown, but may be preceded by upper respiratory tract infection
- Most commonly seen in male children 3–10 years
- Most commonly limited to one side

SIGNS & SYMPTOMS

- May be asymptomatic
- Tenderness/pain during passive movement
- One-sided pain in the hip, groin, thigh, knee
- Antalgic limping

DIAGNOSIS

- Diagnosis of exclusion

DIAGNOSTIC IMAGING

Ultrasound

- Fluids in joint capsule

LAB RESULTS

- Slightly ↑ white blood cell count
- ↑ Erythrocyte sedimentation rate
- ↑ C-reactive protein
- Needle aspiration
  - Differentiation between transient synovitis and septic arthritis

OTHER DIAGNOSTICS

- Limited abduction and internal rotation

TREATMENT

MEDICATIONS

- NSAIDs

OTHER INTERVENTIONS

- Massage
- Rest

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

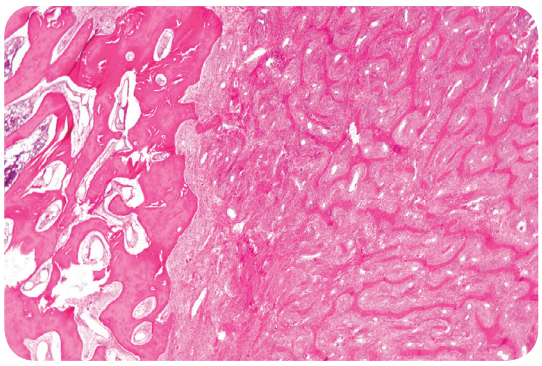

Figure 107.1 The histological appearance of bone in an individual with fibrous dysplasia of bone. Normal bone tissue is on the left. The affected bone is composed of thin disordered trabeculae. The bone marrow spaces are packed with fibrous tissue.

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

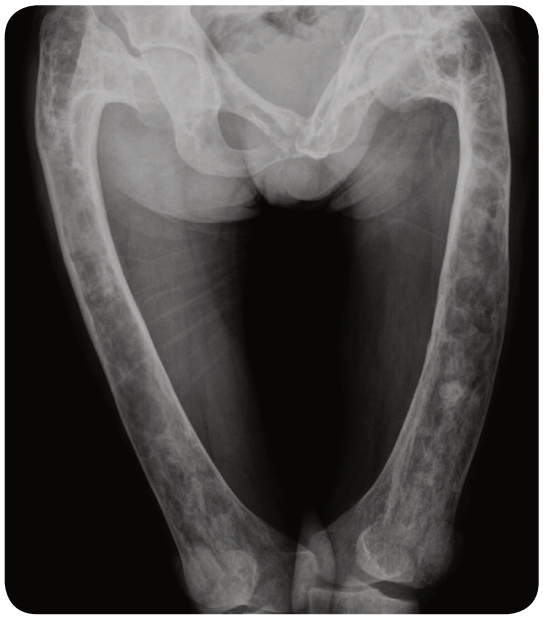

Figure 107.2 An X-ray image of the femurs demonstrating a shepherd's crook malformation. Both femurs are involved in this case of polyostotic fibrous dysplasia.

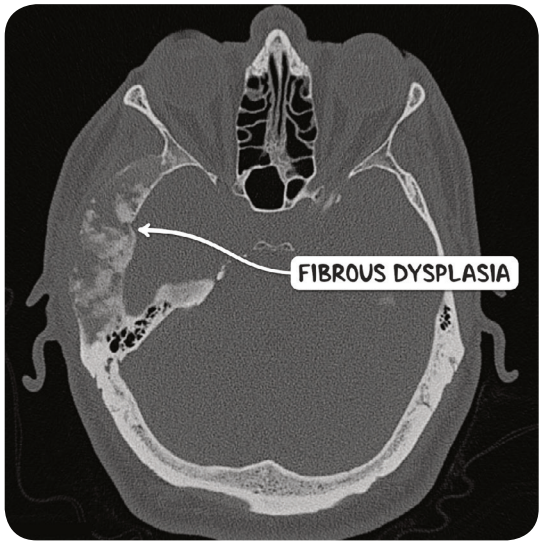

Figure 107.3 A CT scan of the head in the axial plane demonstrating fibrous dysplasia of the squamous temporal bone in a case of monoostotic fibrous dysplasia.

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

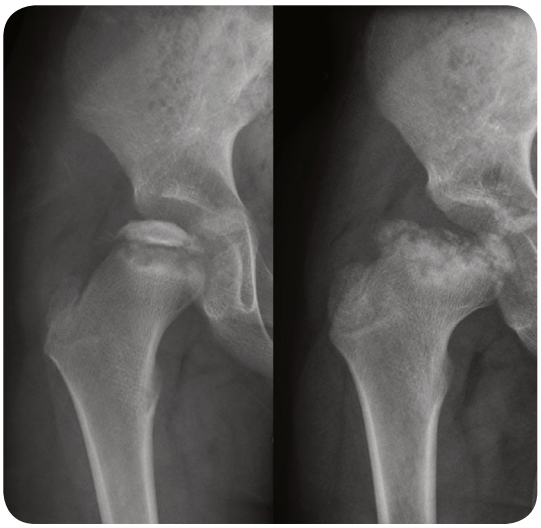

Figure 107.4 X-ray images of the right hip in a child with Legg–Calvé–Perthes disease. The diagnosis was made at six years (left) and by 8.5 years (right) the epiphysis has completely collapsed due to osteonecrosis.

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

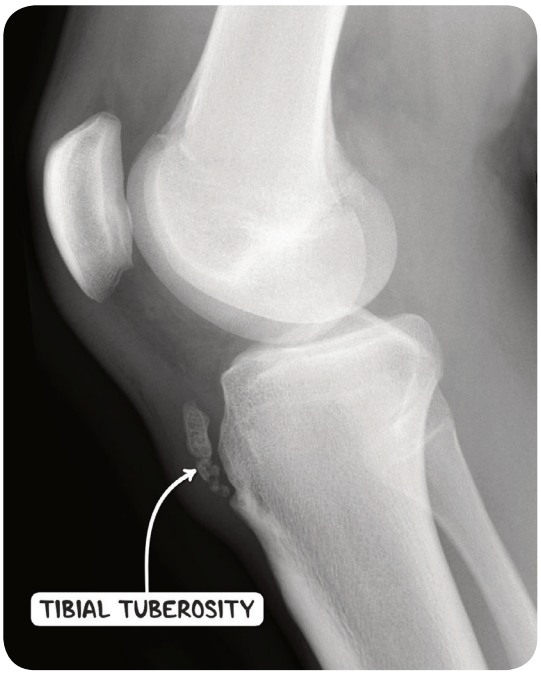

Figure 107.5 A lateral X-ray image of the knee in an individual with Osgood–Schlatter disease. There is fragmentation of the tibial tuberosity and overlying soft tissue swelling.

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

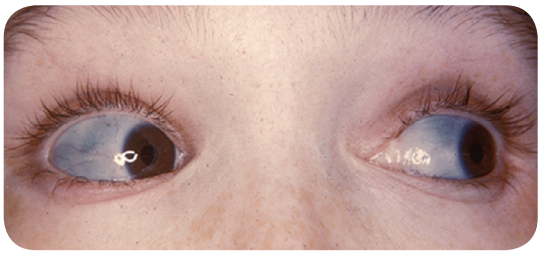

Figure 107.6 Blue sclera in an individual with osteogenesis imperfecta.

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

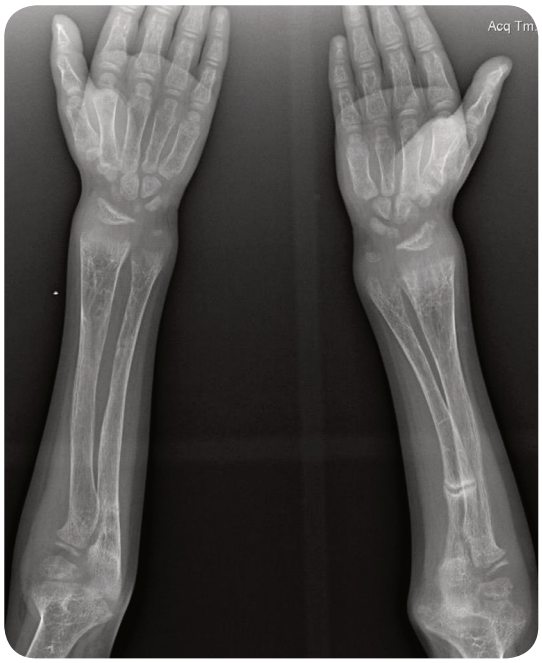

Figure 107.7 X-ray images of the arms of an individual with osteogenesis imperfecta. There is generalised osteoporosis as well as multiple fractures and malformations..

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

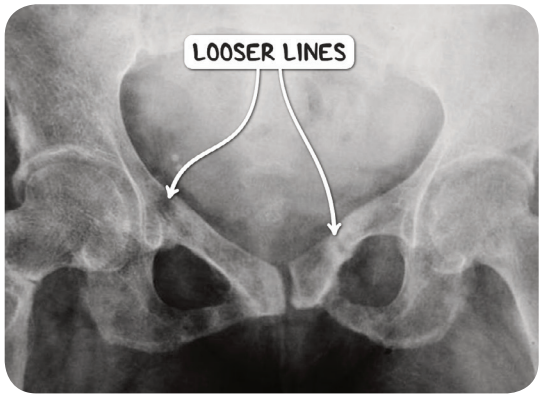

Figure 107.8 An X-ray image of the pelvis of an individual with osteomalacia. There are numerous pseudofractures, or Looser lines, in both the inferior and superior pubic rami.

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

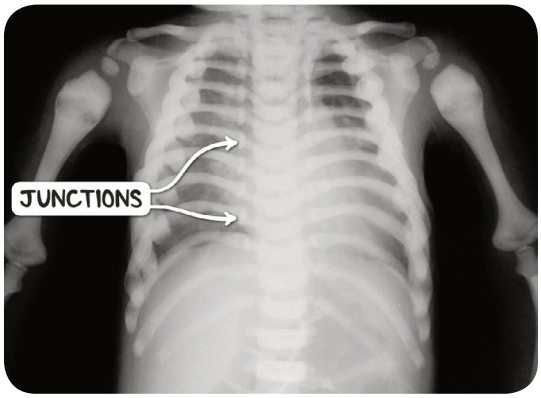

Figure 107.10 A plain chest radiograph of a child with osteopetrosis. There is characteristic widening of the costochondral junctions.

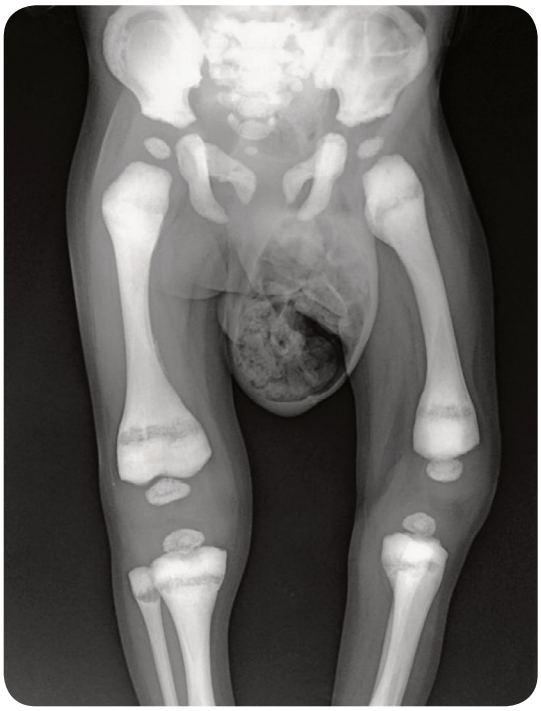

Figure 107.9 An X-ray image of a child with osteopetrosis. There is loss of bony cortico-medullary differentiation and there are lucent metaphyseal bands. There is an incidental large scrotal hernia.

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

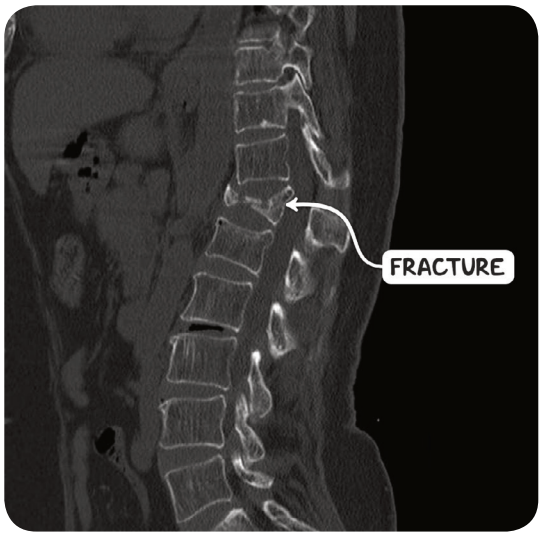

Figure 107.11 An MRI scan of the spine in the sagittal plane demonstrating a compression fracture of T12 secondary to osteoporosis.

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

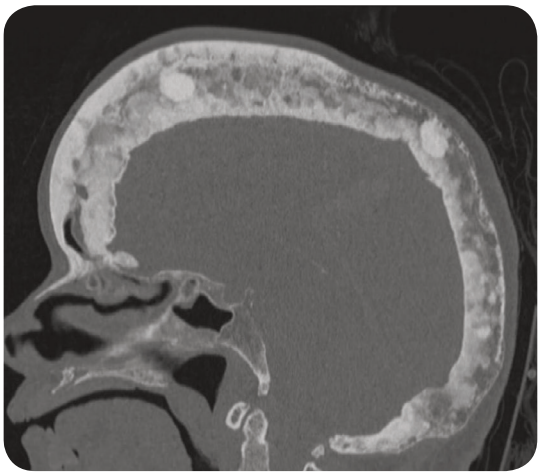

Figure 107.12 A CT scan of the head in the sagittal plane. The skull has the typical cotton wool appearance of Paget's disease of the bone.

- "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

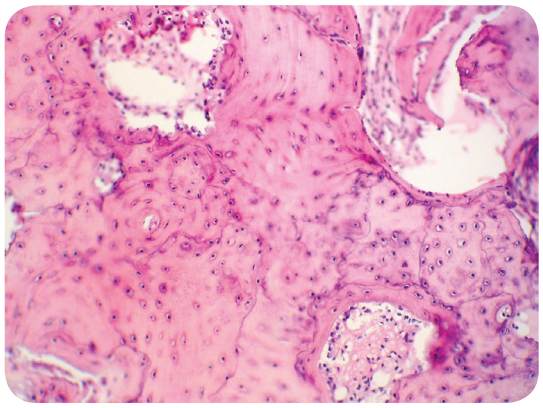

Figure 107.13 The histological appearance of bone in the Paget's disease. Bone formation is increased and highly disordered.

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

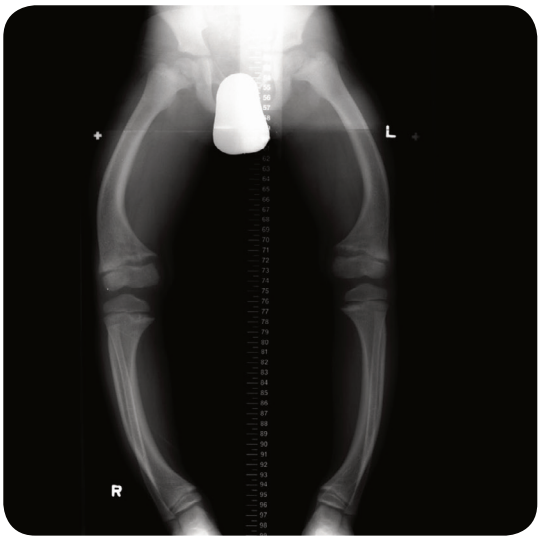

Figure 107.14 An X-ray image of both lower limbs demonstrating bowing in an individual with rickets.

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

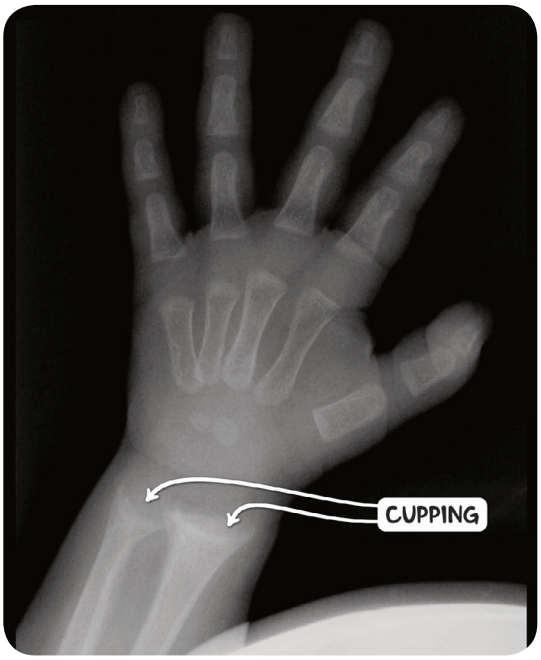

Figure 107.15 An X-ray image of the wrist demonstrating metaphyseal cupping, also known as metaphyseal flaring, in an individual with rickets.

TREATMENT

MEDICATIONS

- Oral vitamin D supplementation
- Correction of calcium intake

OTHER INTERVENTIONS

- Treat underlying causes

NOTES
BONE & JOINT INFECTIONS

GENERALLY, WHAT ARE THEY?

PATHOLOGY & CAUSES

- Bacterial invasion and destruction of bone and joint cartilage
- Caused by bacteria
  - E.g. *Staphylococcus aureus* (*S. aureus*), *Mycobacterium tuberculosis* (*M. tuberculosis*), *Pseudomonas aeruginosa* (*P. aeruginosa*)

RISK FACTORS

- Trauma/open fractures, diabetes/atherosclerosis, orthopedic implants, existing infection etc.

COMPLICATIONS

- Chronic infections
- Bone fractures
- Loss of mobility
- Dissemination of infection

SIGNS & SYMPTOMS

- Pain → individual avoids using infected joint

Systemic

- Fever, chills, weakness, headache

Local

- Swollen, painful, warm

DIAGNOSIS

DIAGNOSTIC IMAGING

MRI

- Detect bone infections

CT scan, X-ray

- Detect fractures

LAB RESULTS

- Blood tests
- *Needle aspiration:* pathogen detection

TREATMENT

MEDICATIONS

- Before identifying pathogen → general antibiotics
- Known pathogen → specific antibiotics

SURGERY

- Surgical cleaning

OSTEOMYELITIS

osms.it/osteomyelitis

PATHOLOGY & CAUSES

- Bacterial infection (*S. aureus*, *M. tuberculosis* most common) → bone, bone marrow inflammation
- Bacteria → bone via bloodstream, nearby infection, open fractures/orthopedic implants
- *First week:* bacterial reproduction → inflammation → bone necrosis (e.g. sequestrum); if periosteum bursts → abscess
- *Later:* cytokines induce bone resorption → replacement with fibrous tissue → new bone formation around necrotic one (e.g. involucrum)

RISK FACTORS

- Diabetes, fractures, splenectomy, orthopedic procedures/hardware

COMPLICATIONS

- If *M. tuberculosis* disseminates from joint to vertebra → Pott disease
- Inadequate treatment → chronic infection → bone fractures, sepsis

SIGNS & SYMPTOMS

Local

- Redness, swelling, painful site, sinus connecting to abscess

Systemic

- Weakness, fever, headache, shivering

DIAGNOSIS

DIAGNOSTIC IMAGING

MRI

- Edema → signal changes
- Cortical loss
- Contrast enhancement of abscess rim

Nuclear medicine scans

- *If MRI not attainable:* higher radiotracer uptake

CT scan, X-ray

- not sensitive for 1–2 weeks
  - Osteopenia (decreased bone density)
  - Periosteal reaction (thickening of periosteum)
  - Aggressive infection → Codman's triangle (lifted periosteum with triangle-shaped, ossified edge)

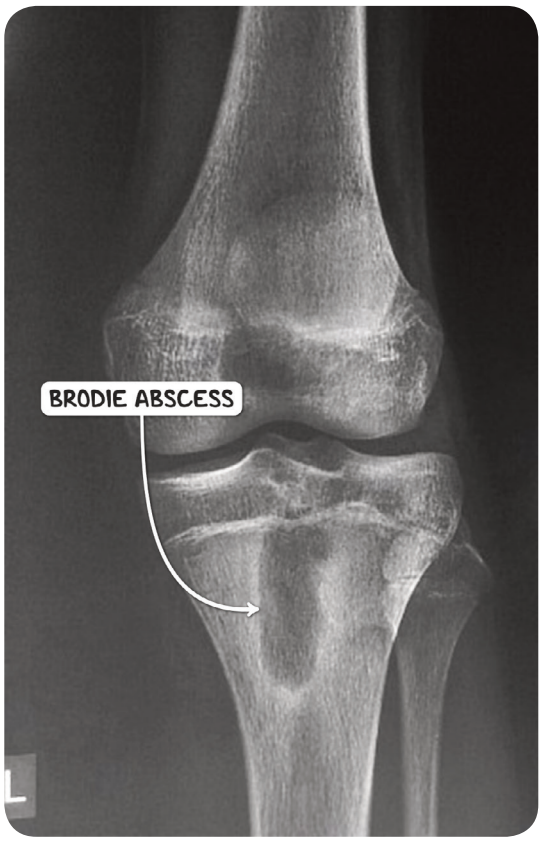

Figure 106.1 An X-ray image of the tibia demonstrating a radiolucency with a sclerotic border consistent with a Brodie abscess.

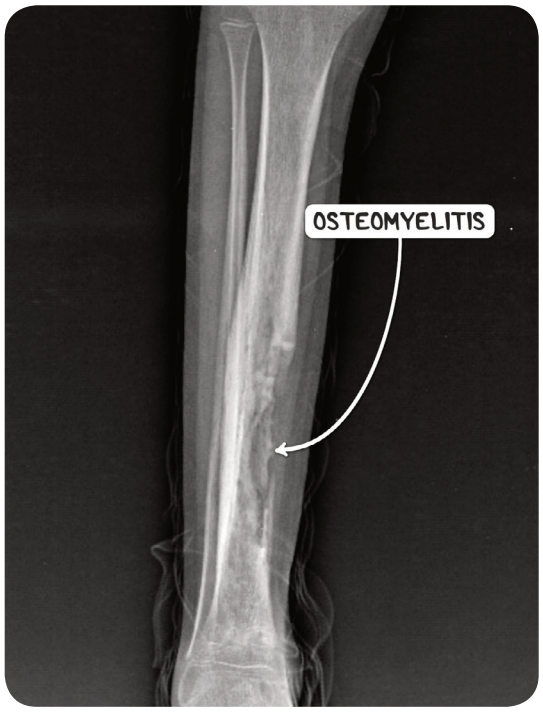

Figure 106.2 A plain radiograph of the right lower leg of an individual with postoperative osteomyelitis. The surgical wound started discharging pus two weeks post open reduction. There is medial cortical destruction and loss of trabeculations with lateral cortical thickening.

- Endosteal scalloping (focal resorption of endosteum)
- Advanced osteomyelitis → cortical bone breakage
- Peripheral sclerosis (increased density at periphery, lower density centrally)

OTHER DIAGNOSTICS

Needle aspiration guided with ultrasound

- Specific antibiotic therapy

TREATMENT

MEDICATIONS

- Long-term intravenous antibiotics

SURGERY

- Surgical removal of dead bone
- Severe cases → amputation

SEPTIC ARTHRITIS

osms.it/septic-arthritis

PATHOLOGY & CAUSES

- *Joint structures:* infected, damaged
- Pathogen enters the joint via bloodstream, from nearby infection/directly (e.g. open fracture)
- Infection of joint → endotoxin production → cytokine release → neutrophil attraction → inflammation, damage of joint structures

CAUSES

- Most commonly *S. aureus* (any age group), *Neisseria gonorrhoeae* (*N. gonorrhoeae*; sexual transmission → adults)

RISK FACTORS

- Diabetes
- Joint trauma
- Artificial joint, surgical procedure
- Osteomyelitis
- Chronic arthritis (e.g. rheumatoid arthritis)
- Immunocompromised
- HIV

SIGNS & SYMPTOMS

- Most commonly affects knee; less commonly ankle, hip, shoulder

Local

- Restricted range of motion; painful, warm, swollen joint

Systemic

- Fever, weakness

DIAGNOSIS

DIAGNOSTIC IMAGING

X-ray, CT scan

- Normal in early stages
- ↑ fluid in synovial part of joint
- Narrowing of joint space due to destruction of cartilage
- Destruction of bone adjacent to cartilage

MRI

- Edema around synovium
- Assess spread of infection outside the joint

Ultrasound

- ↑ fluid
- Guiding needle for aspiration

LAB RESULTS

Blood test

- ↑ white blood cells count (WBC); ↑ sedimentation rate (ESR)

Aspiration of joint fluid

- → bacterial culture → specific antibiotics

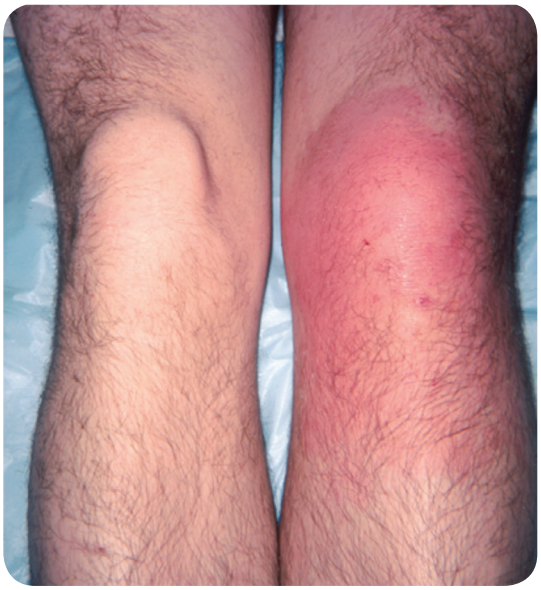

Figure 106.3 A red, hot, swollen left knee in an individual with septic arthritis.

TREATMENT

MEDICATIONS

- General antibiotics depending on Gram stain of joint fluid
- Switch to specific antibiotics once bacteria identified
- Pain medications (e.g. NSAIDs, acetaminophen)

SURGERY

- Surgically drain, cleanse joint fluid

NOTES
HEAD & NECK MUSCULOSKELETAL DISORDERS

GENERALLY, WHAT ARE THEY?

PATHOLOGY & CAUSES

- Disorders of ligaments, muscles, tendons, bones inherent to head, neck

SIGNS & SYMPTOMS

- Most commonly pain

DIAGNOSIS

DIAGNOSTIC IMAGING
- For confirmation

OTHER DIAGNOSTICS
- History, physical examination

TREATMENT

MEDICATIONS
- Anti-inflammatory/muscle relaxant

SURGERY
- In refractory cases

OTHER INTERVENTIONS
- Physical therapy

TEMPOROMANDIBULAR JOINT DYSFUNCTION

osms.it/TMJ-dysfunction

PATHOLOGY & CAUSES

- Category of conditions affecting jaw, producing pain and/or dysfunction centred around temporomandibular joint (TMJ)

CAUSES
- Jaw clenching
- Teeth grinding (bruxism)
    - Nocturnal/diurnal
    - Commonly occurs with MDMA use
- Trauma
    - Reactive oxygen species produced by inflammation → synovial fluid inflammation → cytokine production → TMJ destruction
- Arthritis
- Malocclusion/missing teeth
- Yawning → joint dislocation

- Associated diseases
  - Rheumatoid arthritis (RA)
  - Psychiatric disorders → major depressive disorder

SIGNS & SYMPTOMS

- *Pain:* dull, constant ache; waxing, waning intensity (e.g. headaches, toothaches, earaches)
  - Jaw movement exacerbates (e.g eating, talking)
  - Manifests anywhere trigeminal nerve (cranial nerve V) innervates
- Jaw dysfunction → poor eating/talking ability
- Tinnitus
- Audible popping/clicking of joint

DIAGNOSIS

DIAGNOSTIC IMAGING

Panoramic X-ray

- May reveal frank dislocation of mandible from TMJ

OTHER DIAGNOSTICS

History

- Bruxism
- Trauma

Physical examination

- ↓ Range of motion
- Palpation
  - Tenderness to examiner's finger against TMJ when mouth open
  - Clicking/popping heard/felt when jaw opened/closed
- Abnormal cranial nerve examination
  - Likely trigeminal (CN V) symptom distribution → muscle weakness and/or sensory disturbance

TREATMENT

MEDICATIONS

- Short-term NSAIDs
- Muscle relaxants second line (e.g. cyclobenzaprine)
- *Benzodiazepines:* nocturnal dosing → ↓ nocturnal bruxism

SURGERY

- For refractory disorders
  - Arthroscopy
  - Individuals with underlying arthritis → synovial space bone fragment removal

OTHER INTERVENTIONS

- Pain control
  - Moist heat, cold compresses, massage, soft diet, avoid strain
- Habit adjustment
  - ↓ pen chewing, change sleeping position, oral appliance use
- Physical therapy
- Dislocation → mandible reduction
- Bruxism causative → splinting

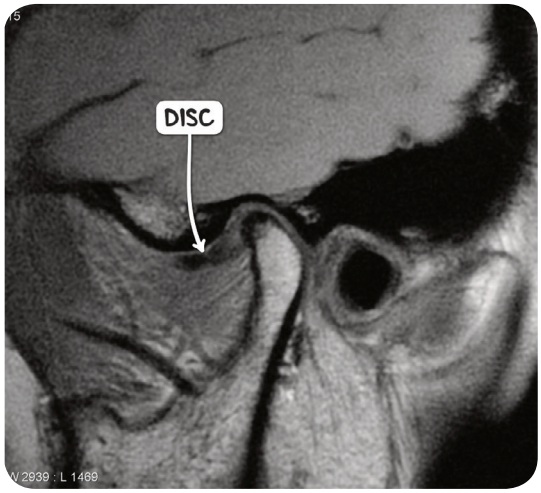

Figure 110.1 An MRI scan of the head in the parasagittal plane demonstrating an anteriorly dislocated disc in an individual reporting symptoms of temporomandibular joint dysfunction.

TORTICOLLIS

osms.it/torticollis

PATHOLOGY & CAUSES

- Abnormal posturing of head, neck; various etiologies
- AKA cervical dystonia
- Sternocleidomastoid (SCM) muscle connect sternums, clavicle (muscle heads) to mastoid process
- One/both SCM head shortened/hypertrophied → contralateral neck flexion, lateral rotation → torticollis

TYPES

Congenital

- Birthing difficulty → injury → fibroma/hematoma formation of SCM muscle → abnormal posturing at/soon after birth
- Spinal abnormalities
- Klippel–Feil syndrome → cervical vertebrae fusion → torticollis
- Atlanto-occipital fusion → abnormal articulation/ankylosis of C1, occipital bone → torticollis

Iatrogenic

- Side effect of dopamine agonist medication (e.g. first-generation antidepressants)

Spasmodic

- AKA adult-onset/idiopathic
- Characterized by tonic/intermittent spasms of cervical muscles in adults

COMPLICATIONS

- Permanent musculoskeletal defects
- Neurologic defects → spinal cord impingement

SIGNS & SYMPTOMS

- Abnormal posturing of the head and neck
  - Lateral rotation (laterocollis)
  - Forward rotation (anterocollis)
  - Backward rotation (retrocollis)
- SCM muscle
  - Hypertrophied
  - Nontender

DIAGNOSIS

OTHER DIAGNOSTICS

- Congenital
  - Birth trauma/condition
- Iatrogenic
  - Coincide with medication schedule/change in dosing
- Spasmodic
  - 5% have ⊕ family history
  - ⅓ have other dystonias

TREATMENT

MEDICATIONS

Congenital

- Muscular etiology → botulinum toxin injections
  - Botulinum toxin → inhibits zinc endopeptidase → inhibition of neurotransmitter vesicle release → decreased muscle contraction → decreased muscle tone

Iatrogenic

- Withdrawal/limitation of offending agent
- Prescription of a muscle relaxant/ antihistamine

Spasmodic

- Muscle relaxant
- Benzodiazepines
- Anticholinergics
  - Side effects → limited use (dry mouth, blurry vision, urinary retention, tachycardia, nausea, vomiting, anxiety)
- Botulinum toxin injections

SURGERY

Congenital

- Vertebral etiology → surgical intervention if severe

Spasmodic

- Refractory cases → surgical denervation of affected cervical musculature

OTHER INTERVENTIONS

Congenital

- Muscular etiology → passive neck stretching

Spasmodic

- Massage
- Physical therapy
- Behavioral modification
- "Sensory trick"
  - Sensory stimulus (e.g. lightly laying hand on cheek) may relieve muscle contraction

NOTES
SPINAL DISORDERS

GENERALLY, WHAT ARE THEY?

PATHOLOGY & CAUSES

- Disorders affecting spinal column
  - Includes vertebrae, intervertebral discs, surrounding structures

RISK FACTORS

- Obesity, extreme exercise/any factor that ↑ spinal column pressure

COMPLICATIONS

- Nerve compression, arthritis, progressive degenerative disease

SIGNS & SYMPTOMS

- Localized pain, stiffness, limited range of motion
- Spine shape irregularities
- Compression → pulmonary, cardiac, gastrointestinal disorders
- Neurologic signs
  - Numbness, paresthesia, weakness, tingling (if nerves affected)

DIAGNOSIS

DIAGNOSTIC IMAGING

MRI

- Detects soft tissue involvement
  - Intervertebral discs, ligaments, nerves

X-ray

- May show osteoarthritis signs
  - Joint pain narrowing, bony spurs

TREATMENT

SURGERY

- If cause irreversible, condition advanced

OTHER INTERVENTIONS

- Malformation
  - Bracing
- Physical rehabilitation, analgesia

DEGENERATIVE DISC DISEASE

osms.it/degenerative-disc-disease

PATHOLOGY & CAUSES

- Progressive intervertebral disc breakdown
- Most common back-pain source
- Accrual of factors → intervertebral disc's nucleus pulposus (mostly water) dehydration → ↓ proteoglycan, collagen → ↓ padding between vertebrae → unable to absorb shock → disc collapse → annular tears, herniation of disc contents into spinal canal → nerve root irritation → nerve impingement → pain

CAUSES

- Multifactorial
  - Accumulation of natural stress, minor injury throughout life
  - Genetic predisposition

RISK FACTORS

- Genetic predisposition, advanced age, menopause, repeated spinal trauma

COMPLICATIONS

- Spine collapse, disc herniation, compression fracture, bony spur growth, neurologic deficit, myelopathy, vertebral artery compression

SIGNS & SYMPTOMS

- Back pain (not correlating to damage's extent), ↓ range of motion
- Pain may radiate
- Tingling, paresthesia, numbness
- Muscle weakness/atrophy
- ↓ deep tendon reflexes
- Headache, dizziness, vertigo

DIAGNOSIS

DIAGNOSTIC IMAGING

MRI

- Evaluates spinal canal, visualizes space available for neural structures
- ↑ signal on T2-weighted images indicate disc dehydration
- Detects annular tears

X-ray

- Detects fracture

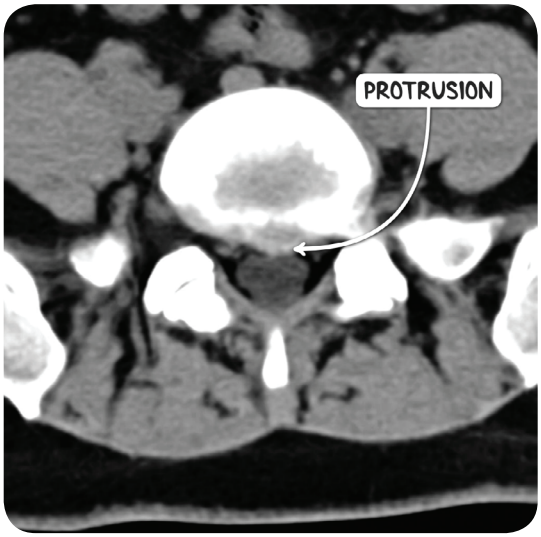

Figure 119.1 An MRI scan of the spine.

TREATMENT

MEDICATIONS

- Pain management
  - Non steroidal anti-inflammatory drugs (NSAIDs), local/epidural corticosteroids

SURGERY

- Corpectomy
  - Vertebral portion removal → enlarge intervertebral space
- Discectomy
  - Herniated disc portion removed
- Nerve root injection
- Intervertebral disc arthroplasty
  - Degenerated discs replaced with artificial discs
- Laminotomy
  - Lamina removal → relieve nerve root pressure

KYPHOSIS

osms.it/kyphosis

PATHOLOGY & CAUSES

- Exaggerated cervical, thoracic, sacral spinal convex curvature
- Greek κυφός *kyphos*, meaning "hump"
- *Cobb angle:* used to measure extent of curvature
  - Angle > 45° classified as kyphosis
- Damage to vertebrae, intervertebral discs/supporting ligaments/muscles → weight-bearing forces asymmetry → further damage to high-pressure area structures → "wedge-shaped" vertebra → spinal curving

TYPES

- May also be caused by trauma/iatrogenic causes (surgery)

Postural

- Most common, occurs all ages

Structural

- Osteoporosis, tumors, tuberculosis (Gibbus malformation), ankylosing spondylitis, fractures, arthritis

Congenital

- Vertebral malformation/*in utero* fusion

Scheuermann's kyphosis

- Adolescent onset, type of osteochondrosis (disordered cartilage ossification)
- Vertebral disc intrudes into end plates in anterior ossification areas (Schmorl's nodes on X-ray)

RISK FACTORS

- Poor posture, weak back muscles, older age, vertebral fracture, osteoporosis, degenerative disc disease, arthritis
- Genetic disease affecting bone, ligaments
  - Osteogenesis imperfecta, Marfan syndrome, Ehler–Danlos syndrome, mucopolysaccharidosis, glycogen storage disease

COMPLICATIONS

- Sternal/vertebral fracture
- Cardiac disease
- Imbalance → fall, fracture risk
- Neurologic
  - Nerve compressions
- Respiratory
  - ↓ pulmonary function
- Gastrointestinal dysfunction
  - Dysphagia, reflux, hernias

SIGNS & SYMPTOMS

- Anterior thoracic pain, dyspnea, limited mobility
- Convex spinal curvature
- Dysphagia, reflux

DIAGNOSIS

- Clinical diagnosis
  - Curvature measurement using flexicurve ruler

DIAGNOSTIC IMAGING

X-ray

- *Sagittal plane:* > 45° Cobb angle
  - Lines drawn above first, last deviating vertebra → draw perpendicular lines to those → angle where they close is Cobb angle

TREATMENT

SURGERY

- Reserved for significant pulmonary/neurologic impairment cases

OTHER INTERVENTIONS

- Milwaukee brace
  - Improves proprioception, helps support back muscles
- Physical therapy strengthens back muscles
- Pain management

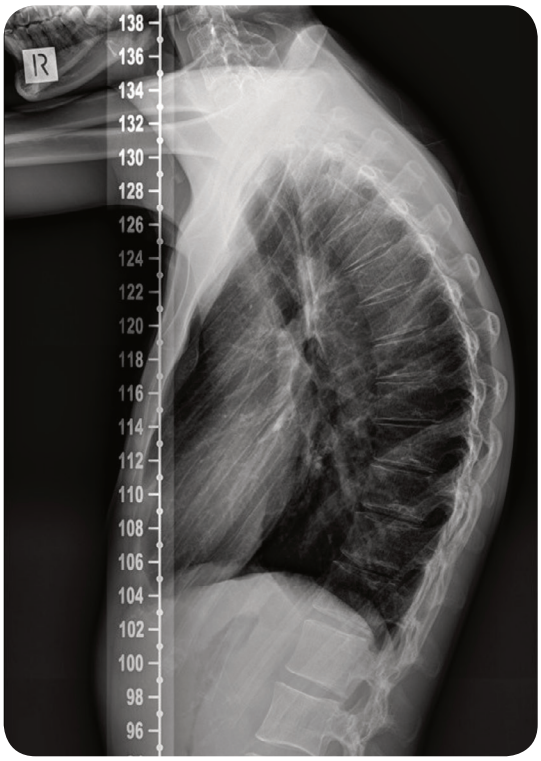

Figure 119.2 A lateral X-ray image of the spine demonstrating marked thoracic kyphosis secondary to Scheuermann's disease.

LORDOSIS

osms.it/lordosis

PATHOLOGY & CAUSES

- Exaggerated inward curvature of lumbar, cervical spine
- Greek *lordōsis*, from *lordos*, meaning "bent backward"
- Bone/neuromuscular imbalance → weight-bearing force asymmetry → further damage to high-pressure area structures/compensatory muscle spasms → spinal curving

CAUSES

- High spine flexibility, lower limb imbalance
- Hip imbalance; improper lifting, squatting
- Muscle strength imbalance (e.g. weak hamstrings; tight hip flexors)
- Obesity
- Osteoporosis
- Spondylolisthesis, discitis
- Temporary lordosis during pregnancy

RISK FACTORS

- Poor posture, muscle strength imbalance
- Musculoskeletal
  - Osteoporosis, spondylolisthesis
- Genetic
  - Achondroplasia, Ehler–Danlos syndrome

COMPLICATIONS

- Degenerative disc disease, nerve compression

SIGNS & SYMPTOMS

- Lower-back pain
- Apparent ↑ lower-back curvature

DIAGNOSIS

- Physical examination reveals ↑ degree of lower-back curvature, muscle tightness

DIAGNOSTIC IMAGING

MRI

- Detects nerve compression

X-ray

- Confirms curvature degree
- Lamina, neural arch of vertebrae may form divert 'V' on anteroposterior lumbar spine radiograph

TREATMENT

OTHER INTERVENTIONS

- Boston brace
  - ↓ disc stress, muscle strengthening
- Physical therapy to strengthen, balance back muscles
- Pain management

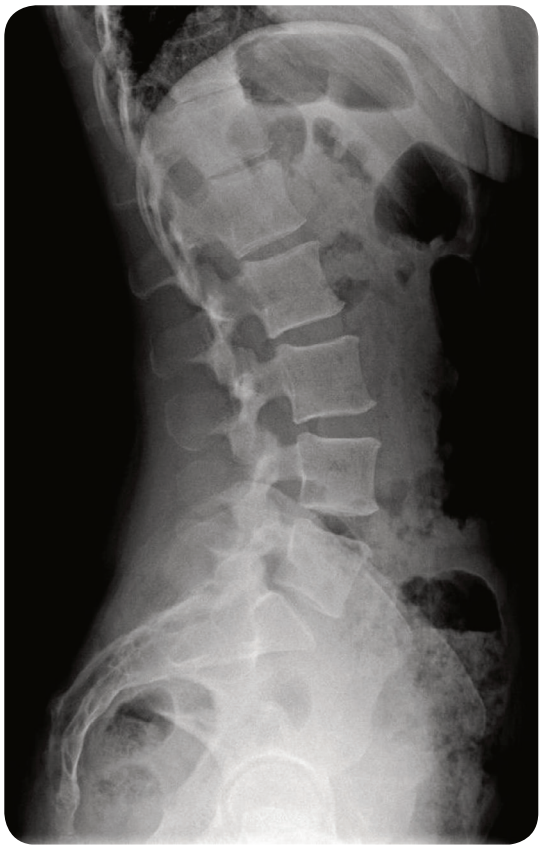

Figure 119.3 A lateral X-ray image of the spine in an individual with hyperlordosis of the lumbar spine.

SCOLIOSIS

osms.it/scoliosis

PATHOLOGY & CAUSES

- Lateral spinal curvature in coronal plane, commonly coexists with rotational curvature
- Bone/neuromuscular imbalance in vertebral/paravertebral area → weight-bearing force asymmetry → further damage to high-pressure area structures → spinal curving
- Sometimes associated with kyphosis, lordosis

Classification according to etiology

- Structural (intrinsic)
- Postural (compensatory)

Classification according to shape

- C/S shaped
- Direction
  - Projection of curvature apex defined with segment involved (most common right-thoracic with left-lumbar presentation)

CAUSES

- Congenital
- Idiopathic
  - Most common; infantile, juvenile, adolescent/early–late onset
  - Multifactorial (environmental, genetic factors)
- Secondary
  - Osteopathic (Marfan syndrome), neuromuscular, neuropathic (neural palsy), myopathic, neurofibromatosis

RISK FACTORS

- Family history
- Obesity
- Lower limb fracture → limb length difference → compensatory scoliosis
- Sudden growth
- Bone tumors, neuromuscular/neural disorders (e.g. Duchenne muscular dystrophy)
- Advanced bone maturity at presentation, biologically-female individuals
  - More severe progression

COMPLICATIONS

- Chest wall abnormalities → respiratory compromise, cardiac complications
- Low self-esteem, depression
- Spinal nerve damage, hemiplegia

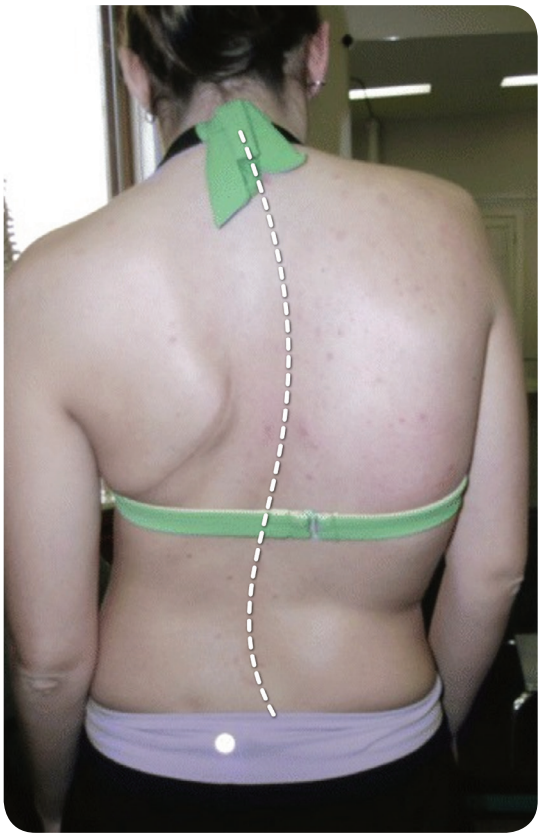

Figure 119.4 An individual with thoracic and lumbar scoliosis. The uneven position of the scapulae is clearly visible.

SIGNS & SYMPTOMS

- Visible spinal curvature, fanning of ribs on one side, uneven musculature
- Back pain
- Difficulty breathing
- Intestinal compression → gastrointestinal difficulty

DIAGNOSIS

DIAGNOSTIC IMAGING

X-ray

- Cobb angle
  - Lines drawn above first, last deviating vertebra → draw perpendicular lines to those → Cobb angle > 10°

OTHER DIAGNOSTICS

Physical examination

- Adams Forward Bend Test
  - Shows torsion, shoulder, pelvis misalignment, unparallel scapulae

TREATMENT

SURGERY

- Cobb angle > 45°
  - Vertebral fusion surgery

OTHER INTERVENTIONS

- Cobb angle < 30°
  - Watchful waiting (frequent check ups estimating curve angle, physical therapy)
- Cobb angle > 30°
  - Boston brace

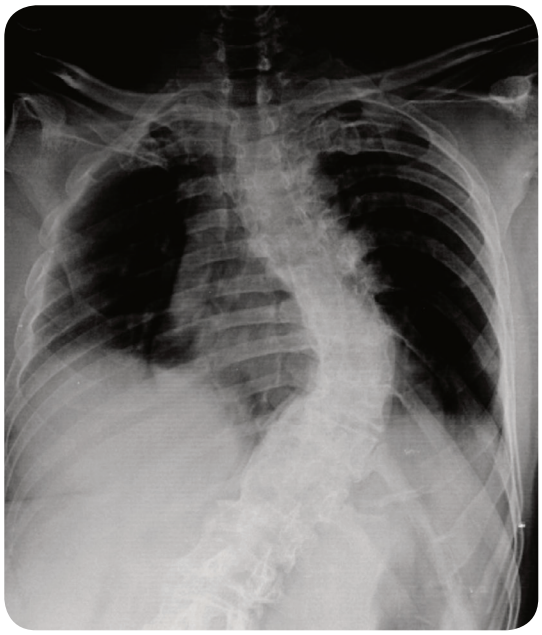

Figure 119.5 A plain chest radiograph demonstrating spinal scoliosis.

SPINAL DISC HERNIATION

osms.it/spinal-disc-herniation

PATHOLOGY & CAUSES

- Middle portion of intervertebral disc (*anulus pulposus*) herniates through tear in outer portion (*anulus fibrosus*) of disc
  - AKA slipped disc
- Weakening of intervertebral disc's outer circle → outer ring tear → inner ring bulging out of spinal column → local nerve compression
- Disc protrusion
  - Outer ring intact but middle portion of disc bulges
  - May → herniation

RISK FACTORS

- Obesity, advanced age, heavy lifting, degenerative disc disease, trauma

COMPLICATIONS

- Nerve impingement
- Sciatica
- Cauda equina syndrome (compression of nerve roots controlling bowel, bladder, legs)

SIGNS & SYMPTOMS

- Continuous pain in certain position, level dependent on injury extent, often unilateral (may present bilaterally)
- Sciatica
  - Lumbar/sacral nerve root compression → pain radiating down legs
- Other symptoms
  - *Sensory:* numbness, paresthesia
  - *Motor:* chronic atrophy, weakness

DIAGNOSIS

DIAGNOSTIC IMAGING

MRI

- Confirm diagnosis

OTHER DIAGNOSTICS

History

- History of heavy lifting, bone degenerative disease

Clinical exam

- Straight leg raise test
  - Individual lies supine, clinician passively raises leg → pain indicates disc herniation

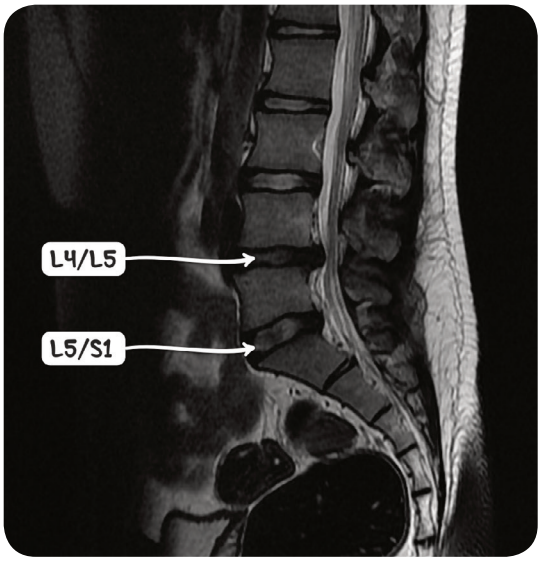

Figure 119.6 An MRI scan of the spine in the sagittal plane demonstrating protrusion of the L4/L5 and L5/S1 intervetebral discs.

TREATMENT

MEDICATIONS

- Pain, inflammation control
  - NSAIDs, local corticosteroids

SURGERY

- Repair
  - In neurologic signs, nerve compression cases

OTHER INTERVENTIONS

- Physical rehabilitation, weight loss

SPINAL STENOSIS

osms.it/spinal-stenosis

PATHOLOGY & CAUSES

- Common chronic condition characterized by narrowing of spinal canal/intervertebral foramina
  - More common in cervical, lumbar regions

CAUSES

- Aging
  - Bone spurs grow into canal, ligaments thicken, slipped discs
- Skeletal disease (e.g. rheumatoid arthritis, osteoarthritis, Paget disease, ankylosing spondylitis, spondylosis, degenerative disc disease)
- Congenital (e.g. achondroplasia, spinal dysraphism)
- Other causes
  - Trauma, fracture, neoplasm, idiopathic

RISK FACTORS

- Obesity, advanced age, family history

COMPLICATIONS

- Cauda equina syndrome
  - Bilateral leg weakness, urinary incontinence
  - Spinal cord narrowing → nerve root compression (L3–S4) → bowel incontinence/sexual dysfunction
  - Neurologic emergency (requires immediate surgical decompression)

SIGNS & SYMPTOMS

- Canal diameter < 10mm
- Neurogenic claudication
  - Discomfort, sensory loss/leg weakness (buttocks, calves)
  - Symptomatic when active (e.g. walking, standing); with lumbar extension
  - Relieved by rest, lying down, waist flexion (squatting, leaning forward)
  - Back pain may coexist
- Radicular pain
  - Pain radiates along dermatome
- Neurologic symptoms
  - Typically bilateral if nerve compression is involved
  - Numbness, weakness, paresthesia, limb pain, urinary/bowel incontinence, sexual dysfunction

DIAGNOSIS

DIAGNOSTIC IMAGING

CT myelogram

- Shows detailed spinal canal contours (if MRI contraindicated)

MRI

- Spinal canal narrowing, nerve compression

TREATMENT

MEDICATIONS

- Pain medications (NSAIDs), epidural steroid injections

SURGERY

- Decompression (advanced disease, complications)

OTHER INTERVENTIONS

- Physical therapy, weight loss

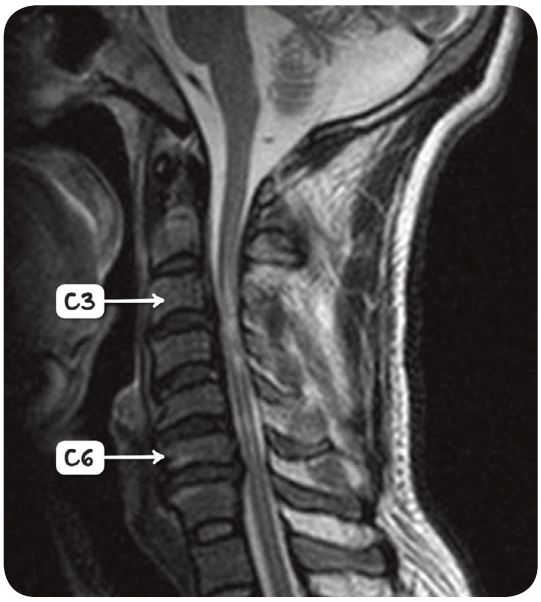

Figure 119.7 An MRI scan of the cervical spine demonstrating spinal stenosis from C3 to C6 resulting in cord compression.

SPONDYLITIS

osms.it/spondylitis

PATHOLOGY & CAUSES

- Chronic vertebrae, vertebral joint inflammation
- AKA spondyloarthritis
- Autoimmune/infectious agent attacks vertebra → inflammatory cells invade site → inflammation, damage to bone, cartilage; thick paravertebral ossification formation
- Spondylodiscitis
  - Vertebrae, intervertebral disc inflammation

CAUSES

- Infectious
- E.g. Pott's disease (osteoarticular tuberculosis); Staphylococcus aureus
- Autoimmune
- Ankylosing spondylitis, rheumatoid arthritis

RISK FACTORS

- Family history
- Immunocompromised state
- Spinal surgery/invasive intervention history

COMPLICATIONS

- Osteoporosis, osteopenia
- Fractures
- Neurologic
- Spinal cord compression, cauda equina syndrome

SIGNS & SYMPTOMS

- Localized pain
  - Segment-dependent
- Gradual symptom onset
  - Autoimmune disease
- Limited movement
- Spinal stiffness
- Symptoms worse in morning, improve with exercise

DIAGNOSIS

DIAGNOSTIC IMAGING

MRI

- Shows calcifications in column; may reveal erosive disease

X-ray

- Asymmetric parasyndesmophytes visualization (paravertebral soft-tissue calcifications)

LAB RESULTS

- Blood tests
  - Infectious cause
- Genetic testing
  - Autoimmune cause

OTHER DIAGNOSTICS

- History of joint pain
- Positive autoimmune disease/exposure history

TREATMENT

MEDICATIONS

- Infectious cause
  - Antibiotics
- Autoimmune disease
  - Disease modifying rheumatoid medications (sulfasalazine, local corticosteroids)
- Pain management (NSAIDS, opioids); severity-dependent

SURGERY

- Severe cases

OTHER INTERVENTIONS

- Physical therapy to strengthen back muscles

SPONDYLOLISTHESIS

osms.it/spondylolisthesis

PATHOLOGY & CAUSES

- Spontaneous anterior/posterior vertebral body slippage over one below it
  - Most commonly affects lumbosacral articulation
- Vertebral joint dysfunction → joint instability → vertebral body slipping from original position

CAUSES

- Lytic/isthmic
  - Most common
  - Multiple *pars interarticularis* microfractures; usually affects athletes
- Degenerative
  - Pseudospondylolisthesis, arthritis, osteoporosis
- Dysplastic
  - Congenital posterior spinal dysplasia elements; usually presents with adolescent growth spurt

- Pathologic
  - Vertebral lesions; neoplastic/infectious infiltration
- Traumatic
  - Facet(s)/pars interarticularis fracture; post-surgical complication

RISK FACTORS

- Genetic predisposition
- Competitive sports (dancing, gymnastics)
- Extreme growth spurt, muscle weakness
- Spinal malformation (Scheuermann's kyphosis)
- Biologically-female individuals

COMPLICATIONS

- Intervertebral disc degeneration
- Spinal stenosis

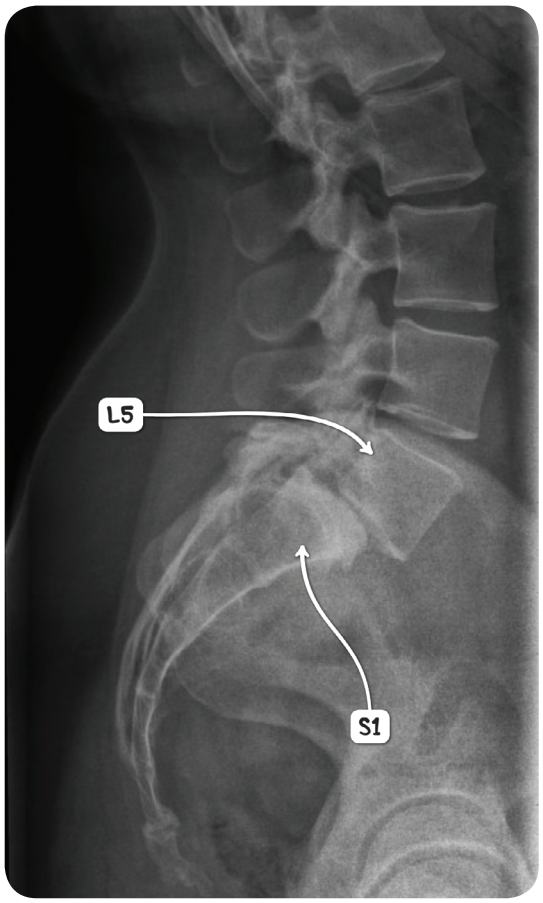

Figure 119.8 A lateral X-ray image of the spine demonstrating spondylolisthesis at L5/S1.

SIGNS & SYMPTOMS

- Chronic back pain/stiffness, posterior leg compartment tightness
  - Pain ↑ with activity, ↓ with rest
- Limited range of motion
- Change in gait (often waddling)
- Forward flexion with development of transverse abdominal crease
- Hip, knee flexion malformations
- Sciatic nerve involvement signs (radiating pain down legs)

DIAGNOSIS

DIAGNOSTIC IMAGING

X-ray or CT scan

- Shows altered vertebral body alignment

TREATMENT

MEDICATIONS

- Pain management (e.g. NSAIDs)

SURGERY

- Repair
  - If persistent pain/neurologic symptoms, vertebral sliding is > 50%

OTHER INTERVENTIONS

- Orthoses (lumbar corset)
  - May help reestablish proprioception, strengthen muscles
- Physical therapy to strengthen back muscles

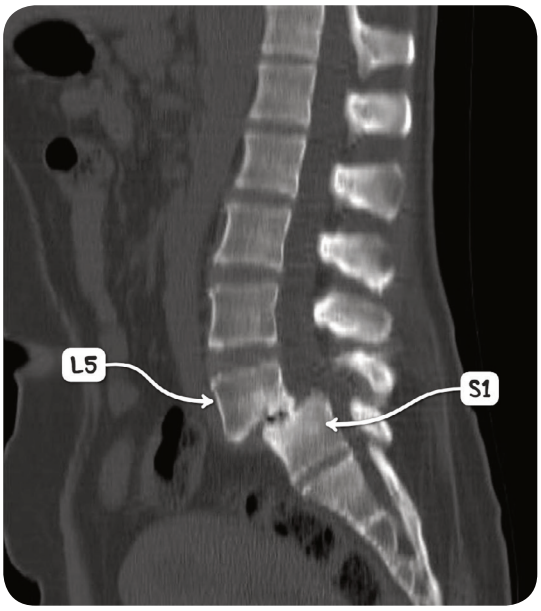

Figure 119.9 A CT scan of the spine in the sagittal plane demonstrating spondylolisthesis of the L5/S1 intervertebral joint.

SPONDYLOLYSIS

osms.it/spondylolysis

PATHOLOGY & CAUSES

- *Pars interarticularis* vertebral defect, mostly lumbar area
  - May be unilateral/bilateral
- Extreme lumbar spine stress → spinal overextension with rotation → vertebral arch fracture/separation

CAUSES

- Unknown, occasionally appears asymptomatically

RISK FACTORS

- Extreme sports during adolescence

COMPLICATIONS

- Sciatica, spondylolisthesis, spinal malformations

SIGNS & SYMPTOMS

- Pain, lumbar spine pressure sensation, focal tenderness
- Unilateral pain radiates into corresponding side's leg
- Painful lumbar spine extension
- Antalgic gait
  - Stance phase of gait shorter than swing phase as means of avoiding pain
- ↑ lumbar lordosis
- Hamstring tightness

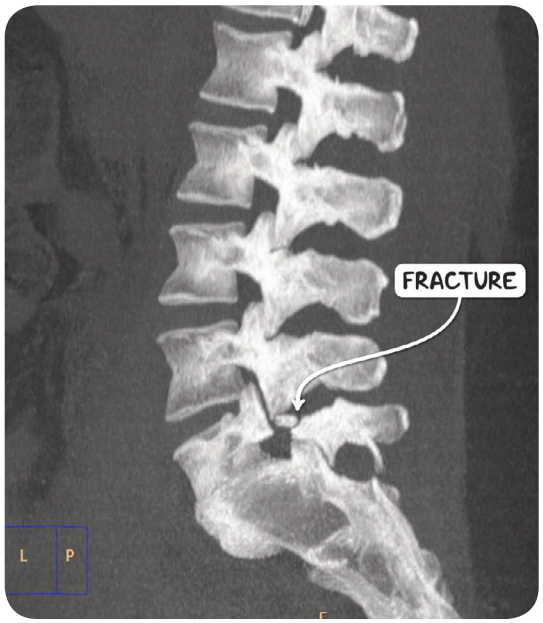

Figure 119.10 A CT scan of the spine in the sagittal plane demonstrating spondylolisthesis of the L5/S1 intervertebral joint.

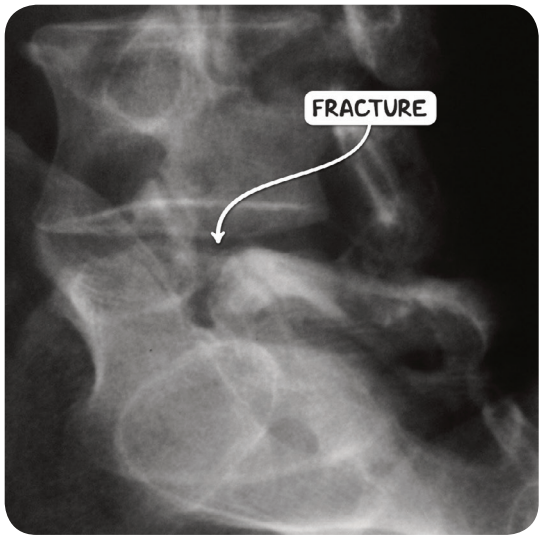

Figure 119.11 A plain radiograph of the spine shows the "scotty dog sign" in a case of spondylolysis.

DIAGNOSIS

DIAGNOSTIC IMAGING

MRI

- Used if neurological findings; visualizes soft tissue, neural structures

X-ray/CT scan

- Lucency in region of *pars interarticularis*
- Description
  - Collar/"broken neck on the Scotty dog" in lateral oblique view

OTHER DIAGNOSTICS

- Clinical exam
  - Stork test (ask to stand on one leg, lift the other hip), tenderness on palpation in fracture area

TREATMENT

MEDICATIONS

- Pain management

OTHER INTERVENTIONS

- Boston brace
- Physical therapy to strengthen back muscles

SPONDYLOSIS

osms.it/spondylosis

PATHOLOGY & CAUSES

- Spinal column degeneration, compression
- Spinal osteoarthritis → degeneration of vertebral bodies, joints, foramina → intervertebral space narrowing → compression, damage to nerve roots

CAUSES

- Osteoartritis, trauma, postural

RISK FACTORS

- Obesity, older age, hyperkyphosis/ hyperlordosis

COMPLICATIONS

- Nerve compression, vertebrobasilar insufficiency, spinal disc protrusion, myelopathy

SIGNS & SYMPTOMS

- Progressive pain in affected spinal region, ↓ range of motion
- If nerves involved
  - Paresthesia, radiating pain, numbness

DIAGNOSIS

DIAGNOSTIC IMAGING

MRI

- Shows nerve impingement and disc abnormalities

OTHER DIANOSTICS

- Clinical exam
  - *Cervical compression test:* lateral flexion of head causes pain in neck, shoulder on same side

TREATMENT

MEDICATIONS

- Pain management

SURGERY

- Alleviate neural impingement

OTHER INTERVENTIONS

- Braces
- Physical therapy to strengthen back muscles

NOTES
UPPER LIMB INJURY

GENERALLY, WHAT IS IT?

PATHOLOGY & CAUSES

- Upper limb joint damage, dislocation
- Weakening/damaging of ligaments, tendons → distortion of normal anatomical joint structure, function loss/impairment

CAUSES

- Trauma, overuse

SIGNS & SYMPTOMS

- Pain, swelling, numbness
- Reduced range of motion
- Visible/palpable malformations

DIAGNOSIS

DIAGNOSTIC IMAGING

X-ray

- Dislocation
- Distorted articular spaces
- Fractures

MRI

- Damaged ligaments and soft tissue

Ultrasound

- Soft tissue damage
- Tendon dislocations

OTHER DIAGNOSTICS

- Physical examination
  - Reduced range of motion
  - Visible dislocation
  - Swelling

TREATMENT

MEDICATIONS

- Pain management
  - Sedation, analgesia

SURGERY

- Joint reduction

OTHER INTERVENTIONS

- Rest, ice
- Physical rehabilitation

DISLOCATED SHOULDER

osms.it/dislocated-shoulder

PATHOLOGY & CAUSES

- Humeral head detaches from glenoid fossa in glenohumeral joint
- Loose ligaments provide high mobility, but are prone to injury in abduction/external rotation

TYPES

Anterior dislocation

- Most common
- Blow to extended, raised, outwardly turned arm → damaged inferior glenohumeral ligament

Posterior dislocation

- Strong muscle cramp/electric shock
- Associated with tuberosity, surgical neck fractures of humerus

Inferior dislocation

- Uncommon
- Force applied to completely raised arm (e.g. individual falls, tries to grab onto something above)
- Highest incidence of axillary nerve, artery injuries

CAUSES

- Force from fall/blow → ligaments tear/stretch → humeral head slips out of position

RISK FACTORS

- Previous dislocations
- Sports

COMPLICATIONS

- Axillary artery, nerve damage from injury/while performing reduction

SIGNS & SYMPTOMS

- Shoulder feels unstable/like it's "rolling out"
- Shoulder pain (can radiate down arm)
- Limited range of motion
- Visible displacement
- *Injured/compressed axillary artery*: hematoma/weak distal pulse
- *Stretched axillary nerve*: shoulder area numbness

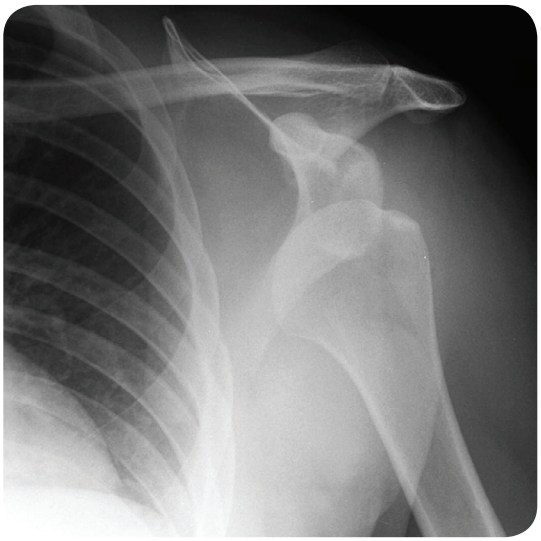

Figure 121.1 An X-ray image of the shoulder demonstrating an anterior dislocation.

DIAGNOSIS

DIAGNOSTIC IMAGING

X-ray

- Two views necessary
  - Anteroposterior view (AP)
  - Lateral view
- *Anterior dislocation*: humeral head is in front of glenoid

- *Posterior dislocation*: humeral head is in place in AP view
- Fractured bones

MRI

- Damaged ligaments (contrast enhancement for better visualisation)

CT angiogram

- Arterial damage

TREATMENT

- Reduction
  - Perform as soon as possible for easier reduction, less chance of complications
  - e.g. Hill–Sachs lesion/compression fractures on humeral head
- Immobilisation, rest
  - Age < 30 → three weeks
  - Age > 30 → 7–10 days

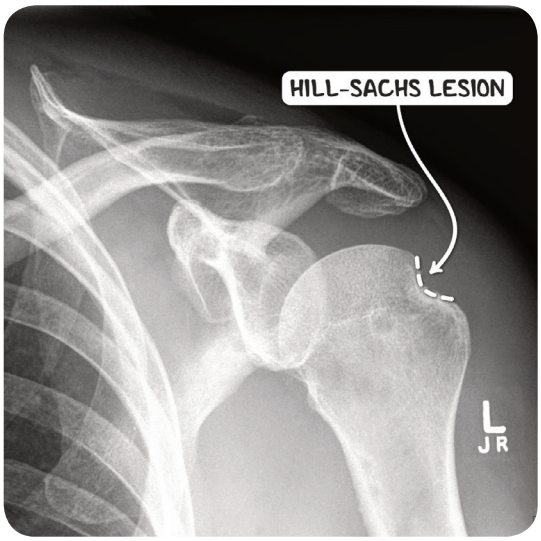

Figure 121.2 An X-ray image of the left shoulder demonstrating a Hill–Sachs lesion. A Hill–Sachs lesion is a posterolateral compression fracture that occurs as a result of recurrent anterior dislocations of the shoulder.

NURSEMAID'S ELBOW

osms.it/nursemaids-elbow

PATHOLOGY & CAUSES

- Dislocation of radial head in elbow joint
  - Pulled elbow/radial head subluxation

CAUSES

- Pulling extended arm makes annular ligament slip above radial head
- Lifting/swinging child
- Common in children < six years old

SIGNS & SYMPTOMS

- Child refuses to bend/use affected arm
  - Fear of pain
- Holds affected arm in prone position, close to body
- Inability to supinate

DIAGNOSIS

DIAGNOSTIC IMAGING

X-ray

- When fracture suspected

TREATMENT

OTHER INTERVENTIONS

- Reduction
  - Supination, elbow flexion
  - Hyperpronation (less painful)

ROTATOR CUFF TEAR

osms.it/rotator-cuff-tear

PATHOLOGY & CAUSES

- One/more tendons of rotator cuff tear(s)
  - Supraspinatus, infraspinatus, teres minor, subscapularis comprise the rotator cuff
- Most common shoulder problem
  - Occurs in all age groups

TYPES

By course

- Acute tears
  - Strong force damages tendons (e.g. rowing, powerlifting)
- Chronic tears
  - Prolonged repetitive motions (especially overhead moves)
  - *Tendon degeneration*: aging, blood supplies worsen
  - Tendons become irritated, inflamed while passing through narrowed gaps

By amount of damage

- Partial thickness tears
  - Damaged supraspinatus tendon
- Full thickness tears
  - Damaged supraspinatus, infraspinatus, subscapularis, biceps tendon

By exterior/interior factors

- Exterior factors
  - Tendon impingement due to curved/hooked *acromion*
- Interior factors
  - Small repetitive injuries over prolonged period → tendon degeneration

SIGNS & SYMPTOMS

- "Arc of pain" (pain while lowering arm)
- Night pain
- Weakness, instability, restricted range of motion
- Compressed nerves → numbness

DIAGNOSIS

DIAGNOSTIC IMAGING

X-ray

- Anteroposterior view
  - Sclerosis, cyst formation, smaller gap between acromion, humerus
- Lateral view
  - State of acromion
- Axillary view
  - Humeral head position

MRI

- Connective tissue visualization
- Size, location of damage
- Swelling

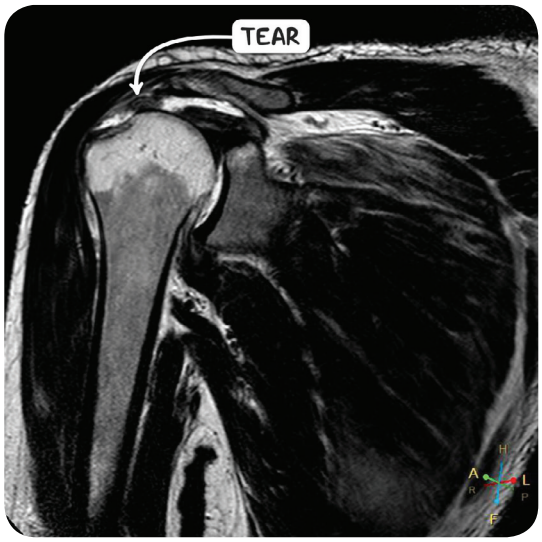

Figure 121.3 An MRI scan of the shoulder in a non-orthogonal plane demonstrating a complete tear of the supraspinatus and infraspinatus tendons.

Ultrasound
- Evaluate tear extent
- Tendon dislocations

OTHER DIAGNOSTICS

Supraspinatus injuries
- Active painful arc & drop arm test
  - Fully raise arm, then steadily lower it back
  - If pain occurs → positive test
- Jobe's test (aka "empty can")
  - Individual raises straight arm 90°, flexes forward 30° with thumb pointing down → resists attempt to depress arm
  - Pain without weakness → tendinopathy
  - Pain with weakness → tendon tear

Infraspinatus, teres minor injuries
- Test external rotation
  - Individual attempts external rotation of arm, examiner provides resistance

Subscapularis injuries
- Gerber's lift-off test
  - Place hand behind back → push backwards against resistance
- Supine Napoleon test
  - Individual lays down, places hand on abdomen with elbow flexed 90° → attempts to raise elbow while examiner secures hand, shoulder

Impingement test
- Neer test
  - Individual flexes pronated arm (with thumb pointing downwards) above head
- Hawkins test
  - Individual raises arm 90° with half-flexed elbow → examiner attempts to internally rotate shoulder

TREATMENT

MEDICATIONS
- Pain management
  - NSAIDs

SURGERY
- Small tears → arthroscopically
- Large tears → open surgical repair

OTHER INTERVENTIONS
- Rest, ice
  - Pain, inflammation management
- Physical therapy
  - Restore range of motion
- Strengthen muscles that support joint
- Exercises for preserving neurologic control

ROTATOR CUFF TEAR OVERVIEW

MUSCLE INJURED	IMPAIRED MOVEMENT	DIAGNOSTIC TEST
SUPRASPINATUS	Abduction	Active painful arc test Drop arm test Jobe's test
INFRASPINATUS	External rotation	External rotation test
TERES MINOR	External rotation	External rotation test
SUBSCAPULARIS	Internal rotation	Gerber's lift-off test Supine Napoleon test

NOTES
LOWER LIMB INJURY

GENERALLY, WHAT IS IT?

PATHOLOGY & CAUSES

- Injury to ligaments, tendons, bony structures of lower extremities

CAUSES

- Trauma, sport

SIGNS & SYMPTOMS

- Pain, swelling in affected region/joint

DIAGNOSIS

DIAGNOSTIC IMAGING

- Imaging to confirm

OTHER DIAGNOSTICS

- *History:* traumatic event, risk factors review
- *Physical examination:* especially provocative (eponymous) musculoskeletal joint evaluation

TREATMENT

MEDICATIONS

Acute

- Analgesics (NSAIDs)

SURGERY

Therapeutic

- Surgical intervention (depending on disability, desire to return to sport/ demanding activity)

OTHER INTERVENTIONS

Acute

- Rest, ice

ACHILLES TENDON RUPTURE

osms.it/achilles-tendon-rupture

PATHOLOGY & CAUSES

- Acute, complete disruption of achilles tendon
  - Commonly traumatic, but can be iatrogenic

CAUSES

- *Recreational Sports:* > 80% of achilles tendon ruptures
  - Increased activity, shear stress on achilles, direct trauma to tendon
  - Sudden, forced dorsiflexion of ankle outside normal range of motion

RISK FACTORS

- *Age:* 30–40 years old
- Biologically-male individuals
- Obesity
- *Fluoroquinolone use:* unknown mechanism
- Systemic corticosteroid use

COMPLICATIONS

- *Re-injury:* 10% of individuals with rupture have history of previous rupture

SIGNS & SYMPTOMS

- Ankle pain
- Poor ambulation

DIAGNOSIS

OTHER DIAGNOSTICS

Physical inspection

- *History:* sudden, painful pop in lower leg; inability to walk; pain immediately after injury
- Calf muscles
  - Soft, lumped together toward knee
- Proximal achilles
  - Likewise collected proximally

Physical examination maneuvers

- Calf squeeze test (Simmonds/Thompson test)
  - Squeezing calf of affected leg does not elicit plantar flexion (very high sensitivity, specificity)
- Palpable gap test
  - Posterior leg palpation at level of achilles to palpate gap in tendon
- Knee flexion test (Matles test)
  - Individual is prone with knees flexed at 90° → observe angle of ankle
  - Ruptured achilles → acute angle (unopposed dorsiflexion of foot by grativity)

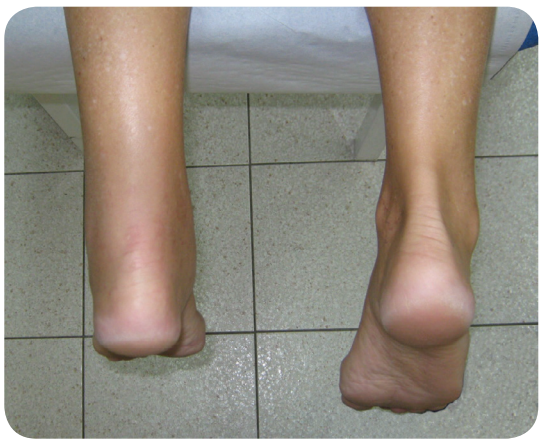

Figure 115.1 A positive Simmond's test (left) in an individual with a ruptured achilles' tendon

TREATMENT

MEDICATIONS

Acute

- Analgesics (NSAIDs/acetaminophen)

SURGERY

Curative

- Orthopedic tendon repair

OTHER INTERVENTIONS

Acute

- Rest, ice

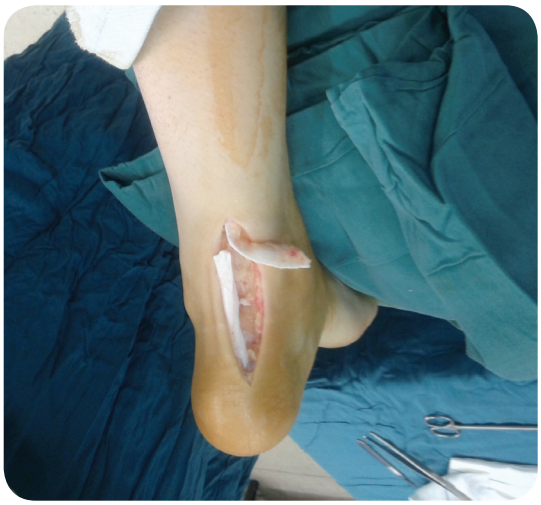

Figure 115.2 A ruptured achilles tendon prior to surgical repair.

ANTERIOR CRUCIATE LIGAMENT INJURY

osms.it/ACL-injury

PATHOLOGY & CAUSES

- Damage/complete tear of anterior cruciate ligament (ACL) in knee; common in deceleration injuries

CAUSES

- *Common mechanism:* twisting knee after planting foot
  - Typically, non-contact injury
  - Common athletic injury

RISK FACTORS

- Biologically-female individuals
- Valgus knee angulation
- ↑ traction ability of field of play
  - *Wet surfaces:* rotation/shift of gravity results in slipping, rather than biomechanical injury to body

COMPLICATIONS

- *Segond fracture:* avulsion fracture of lateral aspect of tibial plateau; occurs in most ACL tears

SIGNS & SYMPTOMS

- Immediate pain
- May have popping sensation/sound at time of injury
- Immediate knee swelling → hemarthrosis
  - Diagnostic maneuvers should be performed immediately after injury for clearest results
- Post-injury
  - Knee may "give out" when walking/standing

DIAGNOSIS

DIAGNOSTIC IMAGING

X-ray

- Rule out fractures (nondiagnostic for ligament tears)

MRI

- Preferred modality to evaluate ligament integrity
- Very high sensitivity, specificity

Knee arthroscopy

OTHER DIAGNOSTICS

Physical inspection

- *History:* pivot sign (knee buckling phenomenon, especially at heel strike phase of walking cycle)
  - Tibia's ability to travel anteriorly (without intact ACL) when knee is flexed at 0–30°→ snaps back around 40°+ of flexion
  - *Underlying this phenomenon:* role of iliotibial band in knee extension, flexion at different degrees of knee position

Physical examination maneuvers

- Anterior drawer test
  - *Supine individual:* affected leg flexes 90°, foot rests on end of bed → examiner sits on foot of affected leg (to stabilize) → grasps around proximal tibia with both hands → pulls anteriorly on tibia → observes anterior movement level
  - *Normal laxity:* < 1cm/0.4in anterior tibial subluxation; negative test, likely intact ACL
  - ↑ *Laxity:* > 1cm/0.4in; positive test, likely torn ACL
- Lachman test
  - *Supine individual:* knee flexed around 20° → examiner flexes knee → grasps around proximal tibia with one hand while stabilizing ipsilateral thigh with other hand → pulls anteriorly on tibia → observes anterior movement level
  - Similar endpoints to anterior drawer test
  - Best sensitivity (85%), specificity (94%) compared to other diagnostic tests

TREATMENT

MEDICATIONS

Acute

- NSAIDs

SURGERY

- Complete ACL tears
- Reconstruction with neighboring patellar ligament/semitendinosus tendon
- *Eligibility:* severity of symptoms, individual's future athletic ambitions
- Majority of individuals elect for surgical repair
- Increased risk of osteoarthritis

OTHER INTERVENTIONS

- *Rehabilitation:* intensive physical therapy

Acute

- Rest, ice, compression of injured knee

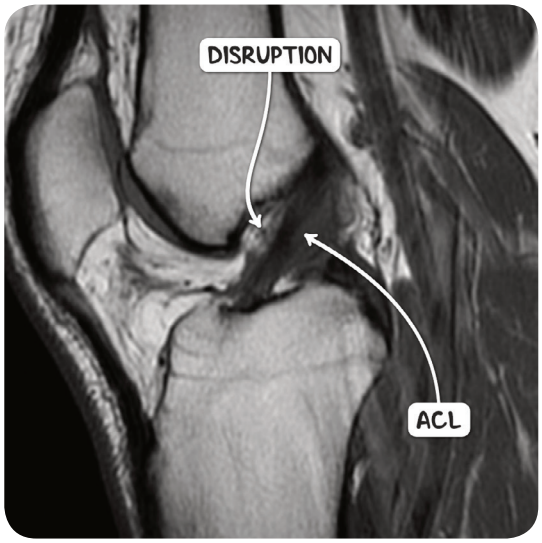

Figure 115.3 An MRI scan of the knee demonstrating partial disruption of the anterior cruciate ligament.

ILIOTIBIAL BAND SYNDROME

osms.it/IT-band-syndrome

PATHOLOGY & CAUSES

- *Painful* overuse *injury*: fibrous band of tissue connects muscles of proximal lower extremity to lateral tibia
- Common injury for runners

CAUSES

- *Iliotibial band (ITB):* involved in knee flexion (at < 30°), knee extension at terminal extension (near 0° flexion); very active in heavy activity → overuse causes inflammation
- Greatest tension across ITB occurs at 30°
  - *Runners:* position of 30° at foot strike → repeat → inflammation, injury
  - *Cyclists:* position of 30° at down-pedal position → repeat → inflammation, injury

RISK FACTORS

Intrinsic

- Weak hip abductors/flexors
- Gastrocnemius, soleus inflexibilty
- Leg length discrepancy

Extrinsic

- Sudden training distance/intensity increase
- *Running:* overstriding, foot eversion (poorly fitted/raised shoes)
- Cold weather exercise

SIGNS & SYMPTOMS

Knee pain

- Sharp/burning, worse during exercise at knee flexion of 30°
- Beyond exercise, pain may ache more/be deeper
- *Location:* lateral femoral epicondyle (LPE)

DIAGNOSIS

OTHER DIAGNOSTICS

Physical inspection

- *History:* Running/cycling with indolent course of lateral knee pain with training

Physical examination maneuvers

- Noble compression test (examiner attempts to recreate pain experienced during training)
  - Individual lays in decubitus position with affected leg above unaffected → examiner puts one thumb proximal to LPE with pressure → examiner uses other hand to passively move affected about the knee from 0–60° flexion → pain → positive test
- Ober test
  - Individual lies on uninvolved side → flexes hip, knee 90° → knee placed in 5° flexion angle → examiner fully abducts lower extremity being tested → allows force of gravity to adduct extremity until hip cannot adduct any further
- Palpation of knee (check for no effusion)
  - Rule out meniscal injury (lateral knee pain, ⊕ effusion)

TREATMENT

MEDICATIONS

Acute

- Analgesics (NSAIDs/acetaminophen)

SURGERY

- *ITB release:* individuals who have failed long-term physical therapy program

OTHER INTERVENTIONS

- Exercise adjustment
  - Address extrinsic risk factors

- Correct leg length discrepancy with insole lift
- Physical therapy
  - Address strength of hip abduction/flexion, calf inflexibility

Acute

- Rest, ice

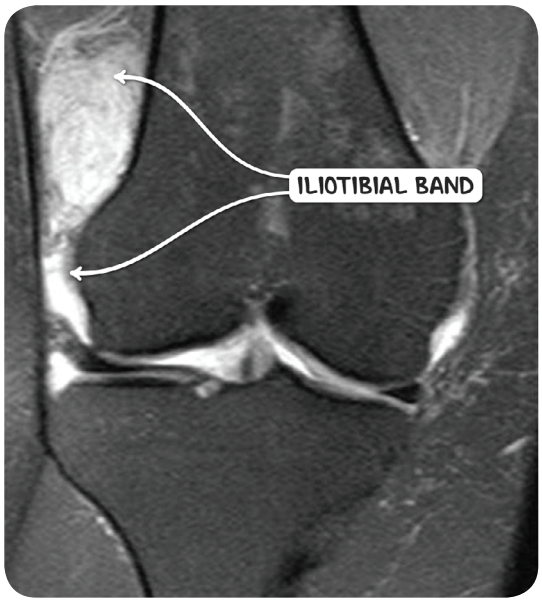

Figure 115.4 An MRI scan in the coronal plane of the right knee of individual complaining of iliotibial band syndrome symptoms. The band is inflammed with surrouding edema close to its point of insertion.

MENISCUS TEAR

osms.it/meniscus-tear

PATHOLOGY & CAUSES

- Injury to fibrocartilage (medial/lateral) knee pads (provide cushion, increase stability at tibiofemoral articulation interface)

CAUSES

- *Pathophysiology:* planted foot → twisting force at knee → compressional, rotational, shear stress placed on meniscus → tear
  - Medial meniscus tears > lateral meniscus tears
  - Medial meniscus firmly attached to medial collateral ligament (MCL) → ↓ mobility of medial meniscus → ↓ force required to tear fibrocartilage
  - Poor blood supply to meniscus via geniculate arteries → poor healing/ regenerative capability post injury

Young, healthy athletes

- Forceful, sudden, decelerating movement while changing direction

Elderly

- Chronic injury requires less torsional force at knee

RISK FACTORS

- Soccer, basketball, American football

COMPLICATIONS

- Osteoarthritis

SIGNS & SYMPTOMS

- Pain at time of injury
- Swelling within 24 hours
- Clicking/crepitus with walking/knee extension
- *Inability to fully extend/lock knee:* occurs in anterior meniscus tears > posterior meniscus tears

DIAGNOSIS

DIAGNOSTIC IMAGING

X-ray

- Non-diagnostic; commonly performed to rule out knee fracture

MRI

- Most sensitive imaging modality for detecting tears
  - *Medial meniscus:* very high sensitivity, specificity
  - *Lateral meniscus:* high sensitivity, very high specificity
- Indicated for surgical evaluation
  - Prevalence of MRI-positive meniscal tears in asymptomatic population increases with age

OTHER DIAGNOSTICS

Physical inspection

- Joint line tenderness (at tibial-femoral interface) because synovial capsule/collateral ligament accompanies injury; less sensitive/specific finding
- Joint effusion likely present

Physical examination maneuvers

- *McMurray test:* tests medial, lateral meniscus
  - Individual is supine with affected knee fully flexed → examiner grasps heel with one hand, around tibial prominence with other hand → exerts rotational force while extending leg → evaluates pain/click/palpable crepitus
  - External rotation → medial meniscus moves under femoral condyle
  - Internal rotation → lateral meniscus moves under femoral condyle
  - Sensitivity (wide range), specificity (high–very high), ⊕ likelihood ratio (LR) (4.0), ⊖ LR (0.6)
- *Apley grinding test:* tests medial meniscus
  - Individual lays in prone position → examiner flexes affected knee to 90° → rotates foot laterally → while stabilizing thigh/femur (with examiner's knee), exerts downward force on tibia
  - *Pain:* likely medial meniscal tear
- *Thessaly test:* tests medial, lateral meniscus
  - Individual stands only on affected leg while holding onto examiner for stability → flexes knee to 20° → rotates knee, body externally/internally
  - *Pain/locking/clicking:* positive test
- *Childress duck-waddle test:* tests posterior horn of medial/lateral meniscus
  - Reserved for athletes fit to complete maneuver
  - Individual squats, walks forward in squatting position → knees are flexed fully → waddling steps exert posterior pressure on knee
  - *Pain/clicking:* positive test

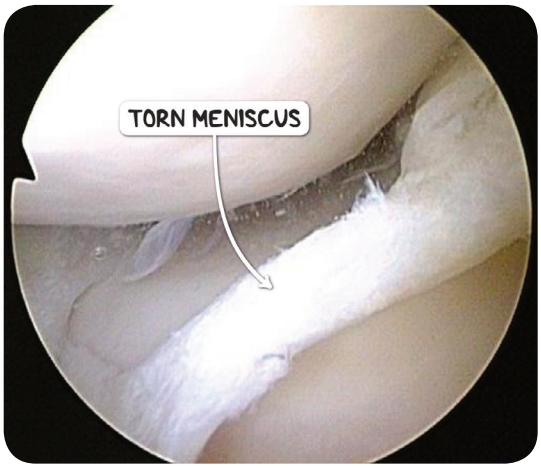

Figure 115.5 An arthroscopic view of a torn medial meniscus.

TREATMENT

SURGERY

- Arthroscopic/open surgery
- Meniscectomy/repair determined by amount of viable tissue intraoperatively, individual's timetable to return to sport/activity
  - *Meniscectomy:* faster timetable to return to baseline activity; long-term ↑ osteoarthritis risk

OTHER INTERVENTIONS

- *Rest:* crutches for severe pain, avoidance of positions/activities that exacerbate pain
- *If knee commonly gives out:* patellar restraining brace; sign of poor quadriceps strength
- Physical therapy

Acute

- Rest, ice

PATELLAR TENDON RUPTURE

osms.it/patellar-tendon-rupture

PATHOLOGY & CAUSES

- Sudden, forced quadriceps contraction against flexed knee, fixed foot

CAUSES

- Most common in individuals < 40 years old involved in heavy training regimens/sport
  - Landing from high jump, making sudden changes in direction at high speed
- *Traumatic injury (non-athletic):* foot/leg is stuck as individual falls backward
  - Body weight falls backward → large eccentric force on fixed leg → force transmitted to patellar tendon → rupture
  - Knee typically fully flexed when injury occurs → exposes tendon to most stress

RISK FACTORS

- Recent glucocorticoid injection
- *Sports with explosive jumping:* basketball, weightlifting
- *Heavy training hours:* > 20 per week
- Biologically-male individuals

COMPLICATIONS

- Tibial tuberosity avulsion fracture
- Patellar fracture/avulsion

SIGNS & SYMPTOMS

- Painful, popping sensation
- Immediate swelling
- *Antalgic gait:* inability to bear weight on affected leg

DIAGNOSIS

DIAGNOSTIC IMAGING

Bedside Ultrasound

- Assists bedside diagnosis

X-ray

- Evaluation of patellar positioning, potential fracture/avulsion complication

MRI

- *Reserved for unusual presentations:* constitutional signs that raise suspicion of tumor

OTHER DIAGNOSTICS

Physical inspection

- *Observation:* upward shift of patella (pathognomonic); swelling distal to patella
- *Strength:* inability to maintain straight leg, raise leg against gravity while supine

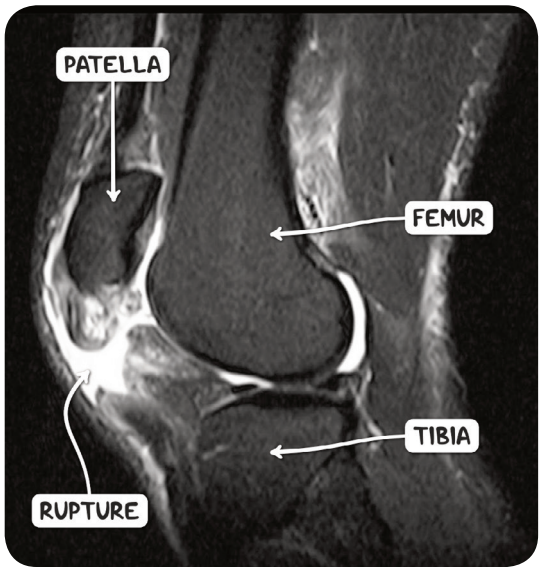

Figure 115.6 An MRI scan of the knee in the sagittal plane demonstrating a patellar tendon rupture

TREATMENT

SURGERY

- Recommended within one week of injury for improved outcomes

OTHER INTERVENTIONS

- Post-operative physical rehabilitation

PATELLOFEMORAL PAIN SYNDROME

osms.it/patellofemoral-pain-syndrome

PATHOLOGY & CAUSES

- Common overuse disorder
- Anterior knee pain that cannot be attributed to intra-articular (meniscus)/peripatellar (patellar tendinopathy) pathology

CAUSES

- Multifactorial pathophysiology

Intrinsic, anatomical factors

- Leg length discrepancy
- Abnormal foot morphology
- Hamstring inflexibility
- Abnormal patellar mobility
- Hallux valgus

Extrinsic, athletic factors

- Exercise overload

RISK FACTORS

- Biologically-female individuals (2:1)
- Active individuals; teens–20s
- Amount of training (ex. distance run) correlates with incidence of disease

SIGNS & SYMPTOMS

- Anterior knee pain
  - Worsened with training, knee extension, especially with squatting, running exercises
  - May occur with prolonged sitting
  - May be present for years
- Knee occasionally buckles/gives way
- *Knee clicks/grinds:* most evident when climbing stairs

DIAGNOSIS

OTHER DIAGNOSTICS

Physical inspection

- History
  - Absence of traumatic inciting event
  - *Achy/sharp knee pain:* typically below patella
- Leg length discrepancy > 1cm/0.4in → poor biomechanics when running → predisposes individual to knee injury
  - < 0.5cm/0.2in leg length discrepancy is common → no increased risk of injury

Physical examination maneuvers

- Palpation
  - Nontender patella, patellar tendon, quadriceps tendon
- Tests
  - *Squatting:* most individuals experience pain
  - *Patellar glide with extended knee:* examiner moves patella laterally
  - Lateral movement ↑ ¾ patellar width abnormal

TREATMENT

MEDICATIONS

Acute

- *NSAIDs (naproxen):* short-term use (2–3 weeks) recommended

OTHER INTERVENTIONS

- Lower extremity muscle strengthening
- *Stretching:* especially hamstrings
- Patellar bracing/taping

Acute

- *Pain control:* avoidance of painful exercise (stair/hill running); substitution of less stressful exercise (stationary bike exercises)

SPRAINED ANKLE

osms.it/sprained-ankle

PATHOLOGY & CAUSES

- Common ankle injury from foot hyper-eversion/inversion

TYPES

Lateral ankle sprain

- Most common
- Inversion of plantar-flexed foot → stretches ankle's lateral ligament complex
- *Lateral ligament:* anterior talofibular ligament (ATFL), calcaneofibular ligament, posterior talofibular ligament

Medial ankle sprain

- Infrequent injury
- Eversion of foot to medial deltoid ligament complex
- Ligament strong enough that medial malleolus fracture is more common than ligament sprain

Syndesmotic sprain

- AKA high ankle sprain
- Injury to interosseous membrane between tibia, fibula
  - Foot is dorsiflexed/ankle eversion
- Higher rate of injury in contact sports (American football)

- Higher rate of chronic ankle sprains → recurrent ankle sprains → ossification of interosseous membrane

SIGNS & SYMPTOMS

- Pain over lateral/medial ankle (depending on eversion/inversion mechanism)
- Swelling hours after inciting event
- Inability to ambulate

DIAGNOSIS

DIAGNOSTIC IMAGING

X-ray

- Evaluate for malleolar, distal fibular, talar dome fracture, syndesmotic separation complication

OTHER DIAGNOSTICS

Physical inspection

- History
  - Mechanism of foot inversion/eversion
  - Prior ankle injuries
  - *Ability to walk after injury:* correlates with fracture complication
- *Observation:* swelling/ecchymosis

Physical examination maneuvers

- Palpation
  - *Fibula, distal tibia:* syndesmotic injury
  - Foot: lateral, medial surface for evaluation of medial, lateral ligament complex pain
  - *Thompson test:* rule out achilles pathology
- Maneuvers of passive inversion/eversion (replicate pain)
  - *Squeeze test (syndesmotic evaluation):* examiner compresses fibular against tibia at level of mid-calf → pain in region of ATFL → likely syndesmotic sprain
  - *External rotation stress test (syndesmotic evaluation):* examiner stabilizes leg proximal to ankle → grasps plantar aspect of foot → externally rotated → pain in region of ATFL → likely syndesmotic sprain
  - *Anterior drawer test (ATFL integrity test):* individual places affected foot in neutral position (slightly plantar-flexed, inverted) → examiner stabilizes lower leg with one hand → grasps heel with other while foot rests on examiner's anterior arm → anterior pull of foot → ↑ laxity of joint (vs. unaffected foot) → likely lateral sprain, ATFL instability
  - *Talar tilt test (calcaneofibular integrity test):* individual places affected foot in neutral position → examiner grasps foot → passive inverts at ankle → ↑ inversion of ankle (vs. unaffected side) → likely lateral sprain, calcaneofibular instability

TREATMENT

MEDICATIONS

Acute

- NSAIDs

SURGERY

- Reserved for ligament rupture in setting of chronic ankle instability

OTHER INTERVENTIONS

- *Rehabilitation:* physical therapy

Acute

- *Rest:* limit weight bearing, use crutches if individual is unable to bear weight
- Ice
- Early application of compressive wrapping → ↓ swelling
- Elevation of ankle → ↓ swelling

UNHAPPY TRIAD

osms.it/unhappy-triad

PATHOLOGY & CAUSES

- Severe knee injury, typically after trauma, that results in trio of ACL, medial collateral ligament (MCL), lateral meniscus tears

CAUSES

- During contact sports, forceful blow to posterolateral aspect of knee, with planted foot (lower body tackle from behind in rugby/American football)
- *Pathophysiology:* posterior force tears ACL → abnormal ↑ anterior glide of tibia relative to femur → medial rotation of tibia → tear of MCL with shearing force → further knee instability → increased rotational force → lateral meniscal tear

COMPLICATIONS

- Osteoarthritis

SIGNS & SYMPTOMS

- Pain
- Hemarthrosis
- Popping/multiple pops

DIAGNOSIS

OTHER DIAGNOSTICS

Physical examination maneuvers

- ACL maneuvers
  - Anterior drawer test
  - Lachman test
- MCL maneuvers
  - Examiner stabilizes affected leg with one hand → exerts valgus stress on lateral aspect of knee → pain and ↑ laxity → likely MCL tear
- Lateral meniscus maneuvers
  - McMurray testing

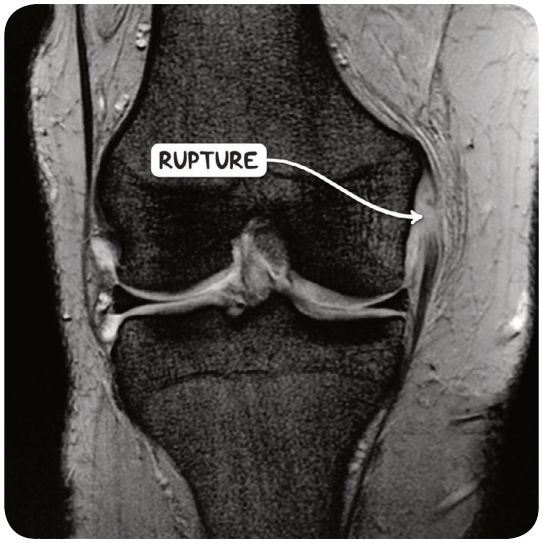

Figure 115.7 An MRI scan of the knee in the coronal plane demonstrating a complete tear of the medial collateral ligament. Injury to the medial collateral ligament is one part of the unhappy triad

TREATMENT

MEDICATIONS

Acute

- NSAIDs

SURGERY

- *ACL repair:* athletes/ individuals who desire return to play/demanding activity
- Lateral meniscus repair vs. meniscectomy

OTHER INTERVENTIONS

Acute

- Rest
- Ice
- Elevation/compression → ↓ swelling
- *Knee brace:* ACL, MCL recovery; overall knee stability

NOTES
TRAUMA COMPLICATIONS

GENERALLY, WHAT ARE THEY?

PATHOLOGY & CAUSES

- Disorders following traumas such as fractures, penetrating trauma, crush injuries, lacerations, hemorrhage, etc.
- Affected tissue damage → tissue homeostasis disruption
  - *Compartment syndrome:* ↑ tissue pressure → blood-flow disruption → tissue ischemia, necrosis
  - *Rhabdomyolysis:* damaged tissue contents release into bloodstream

SIGNS & SYMPTOMS

- Pain, bleeding, tenderness; limb-, life-threatening conditions

DIAGNOSIS

LAB RESULTS

- Complete blood count (CBC), metabolic panel, muscle enzymes, urinalysis

OTHER DIAGNOSTICS

- Asses injury mechanism

TREATMENT

OTHER INTERVENTIONS

- Treat underlying cause

COMPARTMENT SYNDROME

osms.it/compartment-syndrome

PATHOLOGY & CAUSES

- ↑ pressure in closed space compromises space's blood supply
- Any body compartment can be affected (most commonly legs, arms)

TYPES

Acute compartment syndrome

- Most commonly caused by fractures
  - Other causes hemorrhage, crush injuries, vascular puncture, penetrating trauma, severe circumferential burns, intravenous drug injection, revascularization procedures, poor-fitting casts
- Most commonly affects leg, forearm; also foot, thigh, abdomen, gluteal region
- Fracture → hemorrhage, edema, necrotic debris accumulation → ↑ intracompartmental pressure exceeds perfusion pressure (10–30mmHg) → arteriolar collapse → compromised blood flow

Chronic compartment syndrome

- Repetitive muscle use/exertion during vigorous exercise; most commonly affects leg

- Exercise → ↑ muscle size → ↑ intracompartmental pressure → fluid exudation into interstitial space → compromised blood flow

COMPLICATIONS

Acute compartment syndrome

- Untreated → irreversible nerve damage (sensory deficits, paralysis); infection; tissue ischemia → necrosis; limb amputation
- Volkmann's contracture
  - Permanent affected-limb flexion contracture
- Rhabdomyolysis, kidney failure

SIGNS & SYMPTOMS

- Symptom location depends upon affected compartment

Acute compartment syndrome

- Rapid signs, symptoms progression
- Early signs
  - Severe deep pain exacerbated by movement, not relieved by analgesics; swelling, tense muscle compartment, paresthesias
- Late signs
  - ↓ pulse/pulselessness, anesthesia, function loss, paralysis

Chronic compartment syndrome

- Symptoms occur during physical activity, subside when activity stops
  - Insidious pain, tense muscle compartment, numbness, tingling, cramping, muscle bulging, foot-drop

DIAGNOSIS

LAB RESULTS

- Lab findings evident in later stages
- Acute compartment syndrome
  - ↑ creatine kinase (CK)
  - Myoglobinuria

OTHER DIAGNOSTICS

Pressure measurement

- Acute compartment syndrome (usually not required
  - Intracompartmental pressure > 25mmHg
  - Delta pressure < 20–30mmHg (delta pressure = diastolic blood pressure – measured compartment pressure)
- Chronic compartment syndrome
  - Prior to exercise (≥ 15mmHg); after one minute of exercise (≥ 30mmHg); after five minutes of exercise (≥ 20mmHg)
  - Diagnosis of exclusion

TREATMENT

SURGERY

- Acute compartment syndrome
  - *Medical emergency:* immediate management required
  - *Fasciotomy:* all affected compartments → surgical decompression
  - *Escharotomy:* burns
  - *Limb amputation:* severe tissue necrosis
- Chronic compartment syndrome
  - *Fasciotomy/fasciectomy:* moderate–severe cases

OTHER INTERVENTIONS

- Chronic compartment syndrome
  - ↓ exercise volume, physical therapy

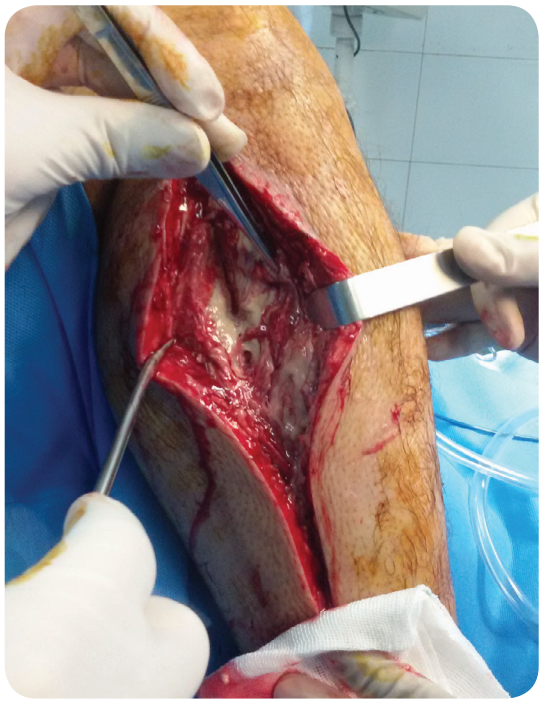

Figure 120.1 Surgeons performing a fasciotomy on the lower leg of in an individual with compartment syndrome.

RHABDOMYOLYSIS

osms.it/rhabdomyolysis

PATHOLOGY & CAUSES

- Extensive muscle necrosis, muscle cell content release into bloodstream
- Muscle damage → ATP depletion → membrane cell pump dysfunction → ↑ intracellular Ca^{2+} due to ↓ efflux → Ca^{2+}-dependent protease activation, hydroxylases, nucleases → cell death, muscle cell content release (myoglobin, CK, Ca^{2+})

CAUSES

- Trauma/compression
  - Crush injury, prolonged immobilization (comatose individuals), acute compartment syndrome, hyperthermia, severe third-degree burns, postoperative surgical trauma
- Metabolic/genetic factors
  - Metabolic myopathies, glycogen storage diseases, mitochondrial disease
- Infections
  - Influenza types A, B; HIV; coxsackievirus
- Medication
  - Statins, colchicine, propofol
- Substance abuse
  - Amphetamines, cocaine, alcohol
- Others
  - Electrolyte disorders, exertional activity, snake venom, hyperkinetic states

COMPLICATIONS

- Acute renal injury
  - Massive myoglobin release exerts toxicity to renal tubules → acute tubular necrosis
- Acute compartment syndrome
- Hyperkalemia, hypocalcemia, metabolic acidosis
- Disseminated intravascular coagulation (DIC)
- Electrolyte imbalances → cardiac arrhythmias

SIGNS & SYMPTOMS

- Severity-dependent
- Non-specific symptoms
  - Fever, nausea, dyspepsia, vomiting
- Muscle pain, weakness, affected muscle swelling
- Oliguria/anuria (acute renal injury)
- Dark urine discoloration

DIAGNOSIS

LAB RESULTS

- ↑ CK
  - Most sensitive, nonspecific indicator
- Myoglobinuria
- Hyperuricemia
  - Blood urea nitrogen (BUN), creatinine
- ↑ Lactate dehydrogenase (LDH)
- Electrolyte imbalances
  - Hyperkalemia, hypocalcemia (early signs)

TREATMENT

OTHER INTERVENTIONS

- IV fluids, electrolyte-imbalance correction
- Kidney function monitoring
- Severe cases
  - Dialysis/hemofiltration

Figure 120.2 Urine of an individual with rhabdomyolysis.

NOTES
BONE TUMORS

GENERALLY, WHAT ARE THEY?

PATHOLOGY & CAUSES

- Abnormal neoplastic bone tissue growth
- Benign tumors more common than malignant

TYPES

- *Primary tumors:* originate from bone (more common)
- *Secondary tumors:* originate from other organs

CAUSES

- Unknown

RISK FACTORS

- Retinoblastoma, Li–Fraumeni syndrome, chronic inflammation, chronic osteomyelitis, Paget's disease, bone infarcts, radiation

SIGNS & SYMPTOMS

- *Usual appearance:* long bones (e.g. femur/tibia), pelvis, vertebra, etc.
- Benign tumors usually asymptomatic, undetected (early stages), discovered by accident
- Pain, swelling, erythema
- Palpable mass, structural malformation
- Pathological fracture, restricted motion range

DIAGNOSIS

DIAGNOSTIC IMAGING

X-ray

- Benign tumors tend to have more defined edges (circular/oval)

CT scan/MRI

- Asses relation with other structures

Nuclear medicine

- Assess skeletal involvement extent, distribution

OTHER DIAGNOSTICS

- Biopsy
  - Histological features

TREATMENT

MEDICATIONS

- Chemotherapy

SURGERY

- Surgery

OTHER INTERVENTIONS

- Radiation therapy

EWING'S SARCOMA

osms.it/ewings-sarcoma

PATHOLOGY & CAUSES

- Rare malignant tumor; bone, soft tissue around bone
- Small, round blue-stained cells; believed to have neuroectodermal origin

CAUSES

- Translocation fuses two regions together
  - *Ewing sarcoma gene (EWS):* chromosome 22
  - *Friend leukemia insertion (FLI1):* chromosome 11
  - *Newly-formed EWS-FLI1 protein:* abnormal transcription factor → induce cell division, malignant transformation

RISK FACTORS

- Usually sporadic; occurs in children, young adults
- ↑ risk, Ewing's sarcoma family history

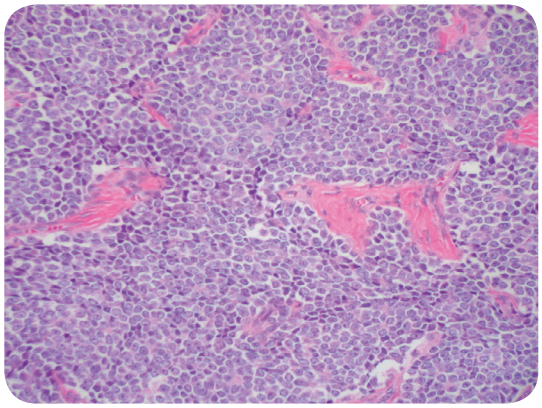

Figure 108.1 A histological section of a classic Ewing's sarcoma. The tumor cells are undifferentiated, closely packed and have vague cytoplasmic borders. There are two distinct populations of light and dark cells.

SIGNS & SYMPTOMS

- Pelvis, femur, clavicle, humerus, ribs, spine commonly affected
- Intense pain (stronger at night); exacerbated by exercise
- Soft palpable mass attached to bone; can compress nerves → function loss (e.g. urinary incontinence if sacrum involved)
- Swelling, erythema
- *Systemic symptoms:* fever, weight loss can indicate metastases
  - *Lung metastases:* most significant cause of death

DIAGNOSIS

DIAGNOSTIC IMAGING

X-ray/CT scan

- Permeative process
  - Bone appears "moth-eaten"
- Periosteal reaction
  - Rapid tumor growth raises periosteum → laminated "onion skin"-like appearance
- Sclerosis

MRI

- Precise tumor location, size, adjacent-structure relation

Positron emission tomography (PET) scan

- Metastases search

OTHER DIAGNOSTICS

- Tumor biopsy
  - Small, round, blue cells

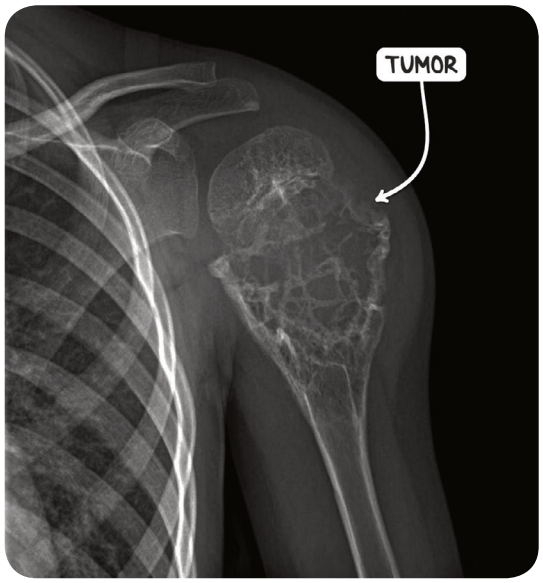

Figure 108.2 An anterior-posterior X-ray of the left shoulder demonstrating a Ewing's sarcoma of the humeral head.

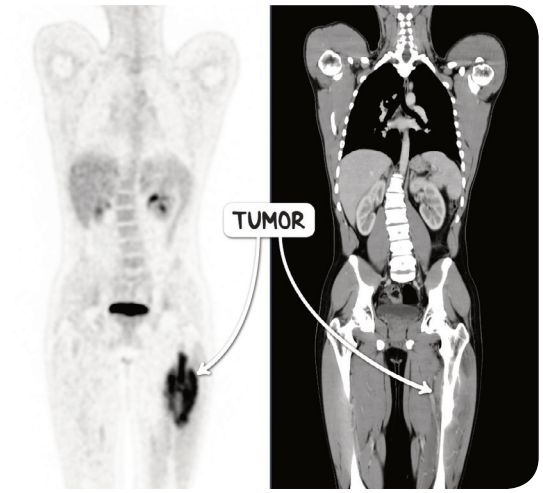

Figure 108.3 A PET-CT scan in the coronal plane demonstrating a Ewing's sarcoma of the left proximal femur. The tumor, visible on CT scan corresponds well with the high levels of tracer uptake on the PET scan.

TREATMENT

MEDICATIONS

- Chemotherapy

SURGERY

- Tumor removal/limb amputation

OTHER INTERVENTIONS

- Radiation therapy

GIANT-CELL TUMOR OF BONE

osms.it/giant-cell_tumor_of_bone

PATHOLOGY & CAUSES

- Benign tumor
  - Destructive growth, metastases potential
- Tumor mass comprises
  - *Tumor cells:* giant multinucleated cells from osteoblastic origin
  - *Non-tumor cells:* osteoclasts, their precursors
- Tumor cells express RANKL → binds to RANK on osteoclasts, precursors membrane → induce cell division, malignant transformation
- ↑ osteoclast number (tumor sometimes called osteoclastoma) + absence of control → bone destruction

SIGNS & SYMPTOMS

- Commonly around knee (distal femur/ proximal tibia)
- Pain, swelling
- Restricted range of motion
- Pathological fractures

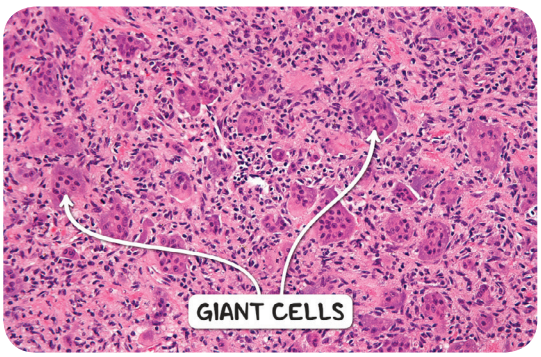

Figure 108.5 The histological appearance of a giant cell tumor of bone. There are numerous multinucleated osteoclastic giant cells surrounded by smaller mononuclear cells.

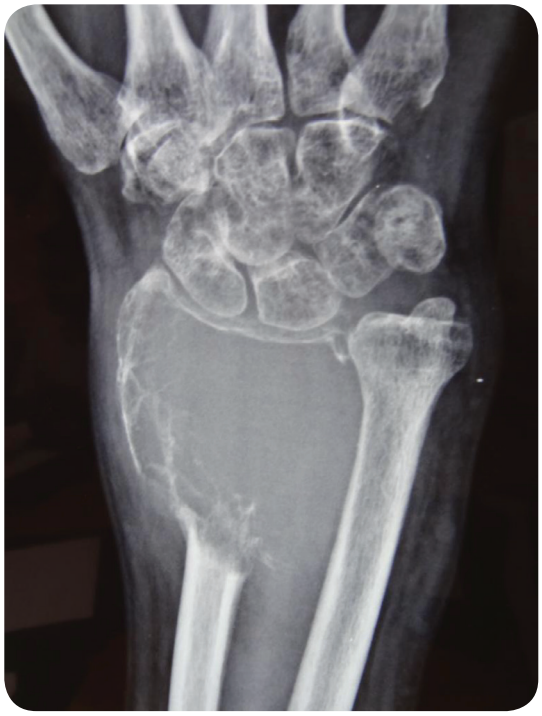

Figure 108.4 A anterior-posterior radiograph of the wrist demonstrating a giant cell tumor of bone at the subarticular portion of the radius. It has a characteristic soap bubble appearance.

DIAGNOSIS

DIAGNOSTIC IMAGING

X-ray/CT scan

- Lucent mass; cortical thinning/destruction; pathological fracture; mineralization, sclerosis absence

MRI

- Homogeneous intensity, well-defined edges

Nuclear medicine

- "Doughnut" sign
  - ↑ periphery uptake, ↓ center uptake

TREATMENT

SURGERY

- Resection

OTHER INTERVENTIONS

- Radiation therapy

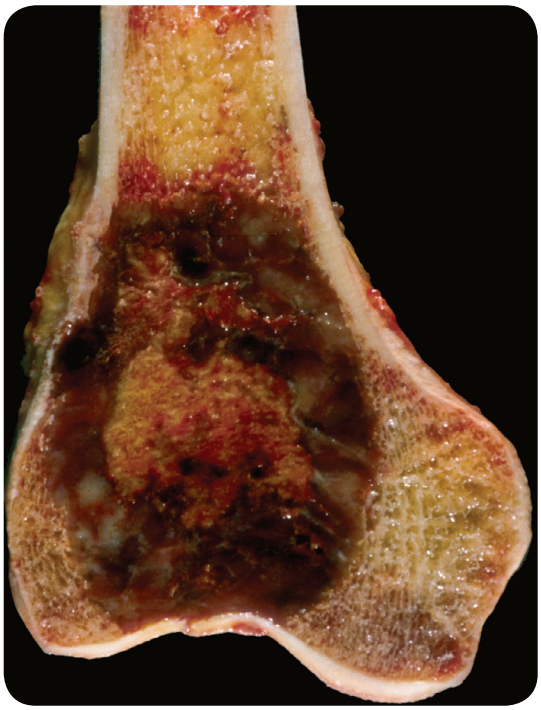

Figure 108.6 The gross pathological appearance of a giant cell tumor of bone affecting the distal femur.

OSTEOBLASTOMA

osms.it/osteoblastoma

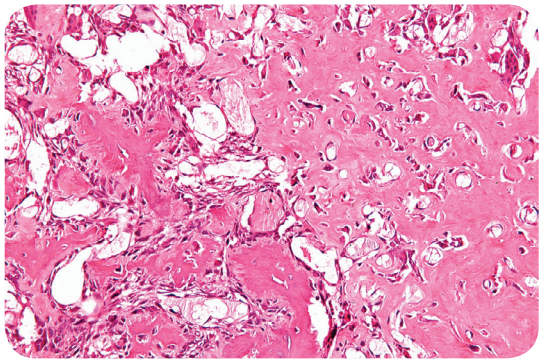

Figure 108.7 The histological appearance of an osteoblastoma. There is abundant osteoid and woven bone forming trabeculae which are lined by a single layer of osteoblasts.

PATHOLOGY & CAUSES

- Benign bone tissue-forming tumor; similar to osteoid osteoma
- Comprises many osteoid (not yet mineralized bone tissue), fibrous (woven) bone-producing osteoblasts
- May break bone cortex, grow to adjacent soft tissue

SIGNS & SYMPTOMS

- Commonly affects posterior spine, long bones (e.g. femur, tibia)
- Dull pain (exacerbated at night)
- Not responsive to salicylates
- Structural malformations (e.g. scoliosis)
- Spinal cord, nerve compression → pain, function loss
- Swelling, tenderness, ↓ range of motion

DIAGNOSIS

DIAGNOSTIC IMAGING

X-ray/CT scan

- Well defined lucent mass
- >2cm/0.79in; no periosteal reaction (differs from osteoid osteoma); if arising from cortical bone → thin new bone layer covering
- Adjacent sclerosis
- Inner calcification

MRI

- Assess surrounding structure volume, relationship

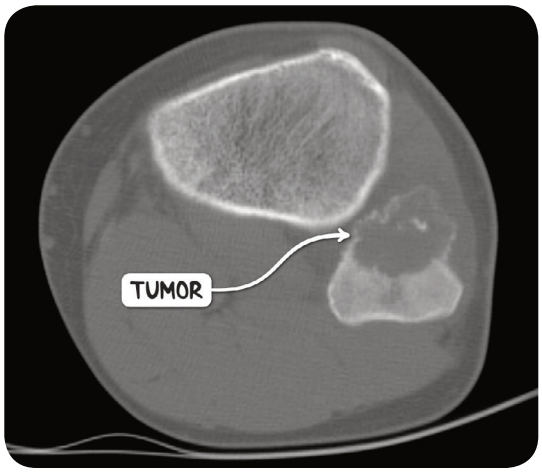

Figure 108.8 A CT scan of the lower leg in the axial plane demonstrating an osteoblastoma of the fibula.

OTHER DIAGNOSTICS

Biopsy

- Immature trabeculae with single osteoblast layer
- High dilated blood vessel number
- Distinguish from osteosarcoma
  - ↓ mitotic activity
  - ↓ cell atypia
  - No cartilaginous matrix
  - Does not imbue surrounding bone, soft tissue

TREATMENT

SURGERY

- Excision

OSTEOID OSTEOMA

osms.it/osteoid-osteoma

PATHOLOGY & CAUSES

- Bone tissue-forming tumor
  - Benign, similar to osteoblastoma
- Tumor mass (nidus)
  - Good blood supply, comprised of osteoblasts providing osteoid, fibrous bone
  - Osteoblasts → ↑ prostaglandin E_2 → pain
- Nidus produces/envelopes itself in reactive bone

SIGNS & SYMPTOMS

- Commonly affects lower-extremity long bones (e.g. femur, tibia), phalanges, spine
- Very intense pain (exacerbated at night)
- Responsive to salicylates
- Painful scoliosis
- Swelling, tenderness

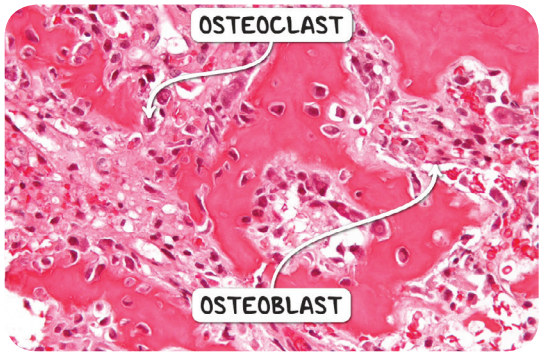

Figure 108.9 The histological appearance of an osteoid osteoma. There is woven bone surrounded by osteoblasts with numerous osteoclasts also present.

DIAGNOSIS

DIAGNOSTIC IMAGING

X-ray/CT scan

- Well-defined lucent nidus
  - Surrounded by reactive bone, <2cm/0.79in in size (differs from osteoblastoma), central mineralization

Scintigraphy

- Higher central uptake

MRI

- Nidus
  - Hard to identify

TREATMENT

MEDICATIONS

- Pain medications (e.g. salicylates, NSAIDs)

SURGERY

- CT scan-guided radiofrequency ablation
- Removal

OSTEOSARCOMA

osms.it/osteosarcoma

PATHOLOGY & CAUSES

- Malignant bone tissue-forming bone tumor
- Growth plates with mutation-prone cells due to proliferation
- Chromosomal aberrations; oncogenes, tumor suppressor mutations
  - *Retinoblastoma gene (RB):* cell cycle regulator
  - *Tumor protein 53 (p53):* DNA repair, cell apoptosis

COMPLICATIONS

- Due to tumor growth
  - Bone marrow repression, cortical bone breakage, adjacent-joint spread

SIGNS & SYMPTOMS

- Appears in long bones, usually around knee (e.g. distal femur, proximal tibia)
- Pain exacerbated by movement; lowered motion range; growing palpable mass; swelling, lymphadenopathy; pathologic fracture

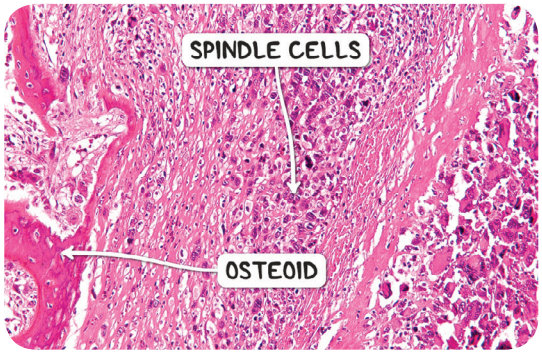

Figure 108.11 The histological appearance of an osteosarcoma. There are numerous malignant spindle cells producing osteoid.

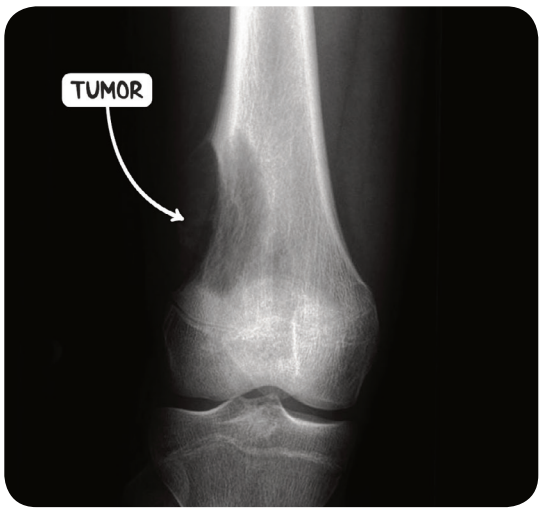

Figure 108.10 An X-ray image of the leg demonstrating a ill-defined, radiolucent lesion at the distal diaphysis and metaphysis of the femur. The tumor was histologically proven to be an osteosarcoma.

DIAGNOSIS

DIAGNOSTIC IMAGING

X-ray

- Soft tissue mass with different calcification range
- Bone destruction
- Codman triangle
  - Periosteal reaction (tumor mass lifts periosteum)
- Permeative process in bone (endosteum appears "moth-eaten")

MRI

- Assess tumor stage, relation to other structures → precise surgical removal

PET scan

- Look for metastases → ↑ radionuclide uptake

OTHER DIAGNOSTICS

- Biopsy, histologic findings
  - Tumor cells produce bone tissue
  - Cell shape/size differs (big cells with hyperchromatic nuclei are dominant)
  - Abnormal mitoses

TREATMENT

MEDICATIONS

- Preoperative chemotherapy (neoadjuvant)

SURGERY

- Surgical removal

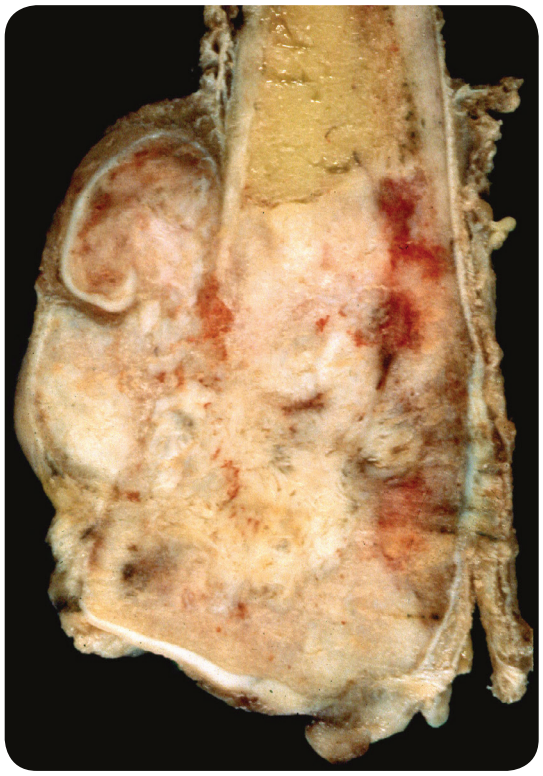

Figure 108.13 The gross pathological appearance of an osteosarcoma of the femur. The tumor is bulky, with a gritty cut surface and cystic degeneration.

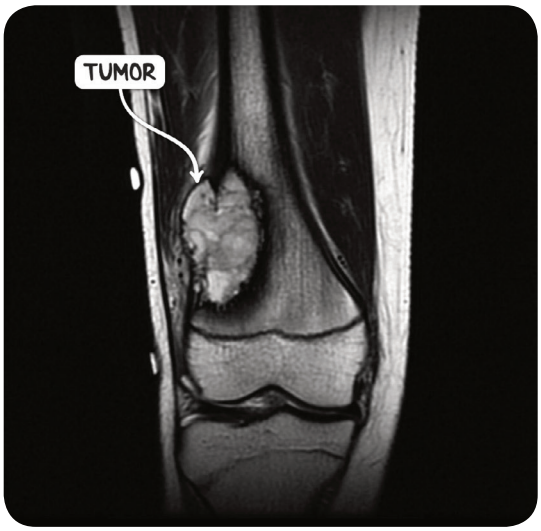

Figure 108.12 An MRI scan of the leg from the same individual. The tumor is seen to extend beyond the cortex of the bone.

NOTES
CARTILAGE TUMORS

GENERALLY, WHAT ARE THEY?

PATHOLOGY & CAUSES

- Tumors arise from cartilage tissue
- Benign/malignant
- Typically located on bones

COMPLICATIONS
- Movement limitation, bone fracture, metastasis

SIGNS & SYMPTOMS

- Can be asymptomatic, enlarging mass, pain

DIAGNOSIS

DIAGNOSTIC IMAGING
- X-ray
- CT scan
- MRI

OTHER DIAGNOSTICS
- Biopsy

TREATMENT

SURGERY
- Resection

CHONDROSARCOMA

osms.it/chondrosarcoma

PATHOLOGY & CAUSES

- Rare bone tumor
  - Endochondral ossification → malignant cartilage production in bones
- Can be primary/secondary, arises from pre-existing benign lesion such as osteochondroma, enchondroma, periosteal chondromas

TYPES
- Conventional (central/medullary), clear cell, mesenchymal, myxoid, dedifferentiated, periosteal (juxtacortical), synovial, extraskeletal
- Affects appendicular skeleton more than axial skeleton

CAUSES
- Unknown

RISK FACTORS
- Age > 50
- History of Ollier disease, Maffucci syndrome, Wilms' tumor, radiotherapy (rare)

COMPLICATIONS
- Pathological bone fracture
- Metastasis
  - Most commonly lungs, bones
- Neurovascular structure impingement
  - Ischemia, venous thrombosis, pseudoaneurysm

SIGNS & SYMPTOMS

- Painful, progressively enlarging mass; localized swelling; limited range of motion; fatigue; weight loss
- Neurovascular involvement
  - Numbness, weakness, skin discoloration, loss of pulse, claudication

DIAGNOSIS

Staging

- Based on grade, spread
  - Intracompartmental, extracompartmental, systemic/regional metastasis

DIAGNOSTIC IMAGING

CT scan

- Matrix calcification, endosteal scalloping

MRI

- *T1:* low–intermediate intensity mass
- *T2:* high intensity mass

X-ray

- *Lytic pattern:* calcifications, endosteal scalloping

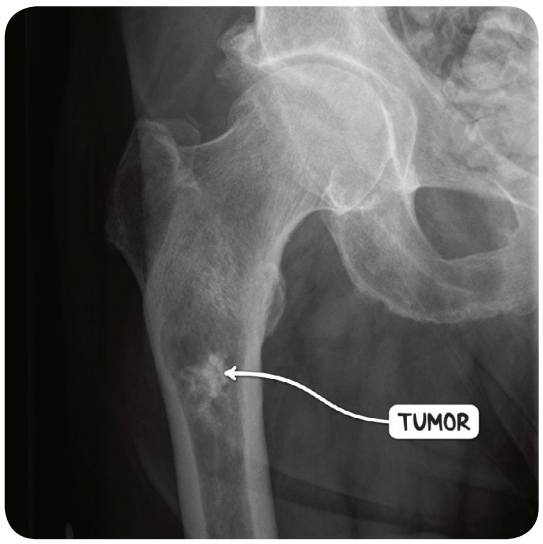

Figure 109.1 An X-ray of an intramedullary lesion with features of a low-grade chondroid lesion, likely a chondrosarcoma.

OTHER DIAGNOSTICS

Fine needle/core biopsy

- Determines histologic grading
  - *Grade 1:* moderately cellular; small, round chondrocyte nuclei; abundant hyaline cartilage matrix; absent mitosis
  - *Grade 2:* ↑ cellularity; ↓ chondroid matrix; enlarged chondrocyte nuclei; scattered mitosis evidence
  - *Grade 3:* ↑ ↑ cellularity, sparse/absent chondroid matrix; nuclear pleomorphism; mitosis clearly present

TREATMENT

SURGERY

- Intralesional curettage + local phenolization/cryotherapy → cementation/bone grafting
- Complete resection

OSTEOCHONDROMA

osms.it/osteochondroma

PATHOLOGY & CAUSES

- Benign tumor; outgrowth of tubular bone growth plate
  - Most common benign bone tumor
  - Average onset age is 10 years
  - Capped with hyaline cartilage
  - Can be pedunculated (with stalk)/sessile (broad base without stalk)
- *Most common localizations:* knee (distal femur/proximal tibia), pelvis, scapula

TYPES

- Single sporadic mass
  - Exostosis
- Multiple tumors
  - Condition known as multiple osteochondromatosis

CAUSES

- Mutation of *EXT1*/*EXT2* genes involved in heparan sulfate glycosaminoglycan synthesis → local glycosaminoglycan reduction → disruption of cartilage, normal skeletal growth
- Radiation-induced
- Idiopathic

COMPLICATIONS

- Pathologic fracture, bone malformation, bursitis, malignant transformation (more common in multiple osteochondromatosis)
- Neurovascular structure impingement
  - Ischemia, venous thrombosis, pseudoaneurysm

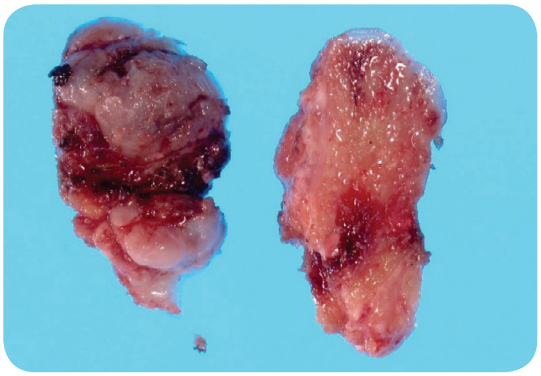

Figure 109.2 The gross pathology of an osteochondroma. The surface of the tumor is composed of hyaline cartilage and the centre composed of cancellous bone.

SIGNS & SYMPTOMS

- Slow-growing palpable mass, pain, impaired range of motion
- Neurovascular involvement
  - Numbness, weakness, skin discoloration, loss of pulse, claudication

DIAGNOSIS

DIAGNOSTIC IMAGING

- Often found incidentally (e.g. radiographic exam performed for different reason)

CT scan

- Evidence of bony lesion and calcification

MRI

- Further characterizes tumor morphology, cartilage cap thickness (thick cap → suspect malignancy)

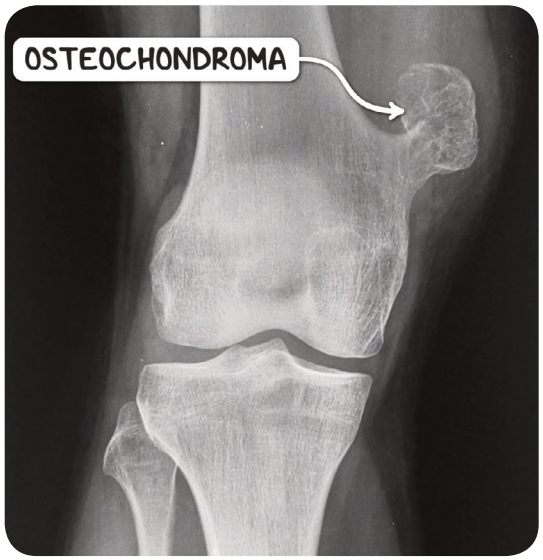

Figure 109.3 An X-ray image of the knee demonstrating a tumor with a cortex continual with normal bone, a characteristic feature of an osteochondroma.

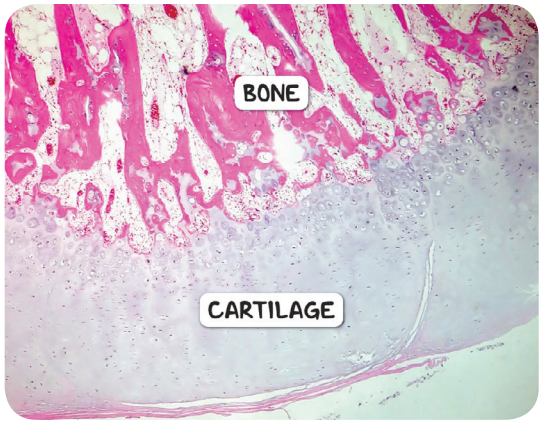

Figure 109.4 The histological appearance of an osteochondroma. There is a core of cancellous bone with an overlying cap of hyaline cartilage.

Ultrasound
- Identifies pseudoaneurysms, thrombosis, bursitis

X-ray
- Dense bony spur

TREATMENT

SURGERY
- Excision
  - Symptoms occur/malignant progression signs

OTHER INTERVENTIONS
- Radiological follow-up
  - Asymptomatic

NOTES
MUSCLE TUMORS

RHABDOMYOSARCOMA (RMS)

osms.it/rhabdomyosarcoma

PATHOLOGY & CAUSES

- Highly malignant tumor arising from skeletal muscle cells

TYPES

Alveolar

Embrional

- Most common, frequently children
  - Botryoid, spindle-cell rhabdomyosarcoma

Pleomorphic (anaplastic)

- Worst outcome, frequently adults

CAUSES

- Genetic translocation determining fusion of two genes which encode two transcription factors
  - FOXO1, PAX3
- FOXO1-PAX3 fusion protein interferes with differentiation-driven gene expression program → poorer outcome

RISK FACTORS

- Syndromes, congenital anomalies (e.g. Li–Fraumeni syndrome, neurofibromatosis type I, Beckwith–Wiedemann syndrome, Costello syndrome)

COMPLICATIONS

- Metastasis
- Bone marrow involvement: anemia, thrombocytopenia, neutropenia

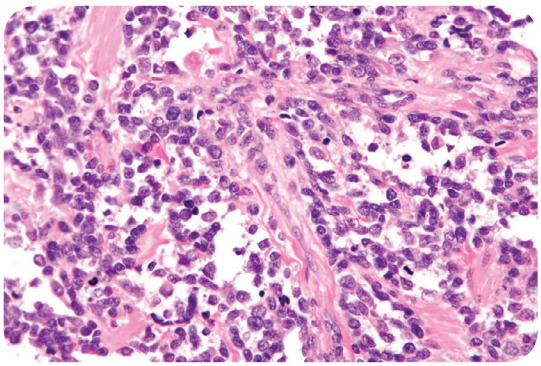

Figure 116.1 The histological appearance of a rhabdomyosarcoma, alveolar subtype. The cells are small, with little cytoplasm with bland chromatin structures visible.

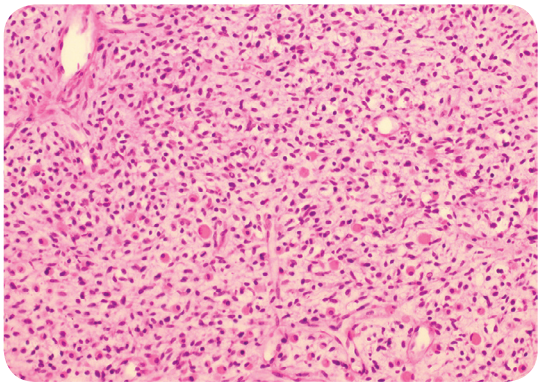

Figure 116.2 The histological appearance of a rhabdomyosarcoma, embryonal subtype. The tumor is composed of numerous round or spindled cells, either with minimal or abundant eosinophilic cytoplasm.

SIGNS & SYMPTOMS

- Palpable mass
  - *Localization:* head/neck, genitourinary tract, limbs, abdomen
- Depending on tumor size
  - Hematuria, urinary obstruction (genitourinary tract involvement)
  - Sinusitis, headache, cranial nerve alterations, ear discharge, orbital swelling (head involvement)

DIAGNOSIS

DIAGNOSTIC IMAGING

X-ray, CT scan, MRI, ultrasound

- Determines local invasion, metastasis

LAB RESULTS

- Liver, renal function tests
- Complete blood count
  - Anemia, pancytopenia
- Biopsy
  - Microscopic analysis
  - Immunohistochemical studies showing muscle tissue-characteristic proteins
  - Genetic sequencing

TREATMENT

MEDICATIONS

- Polychemotherapy
- Immunotherapy

SURGERY

- Resection

OTHER INTERVENTIONS

- Radiation therapy

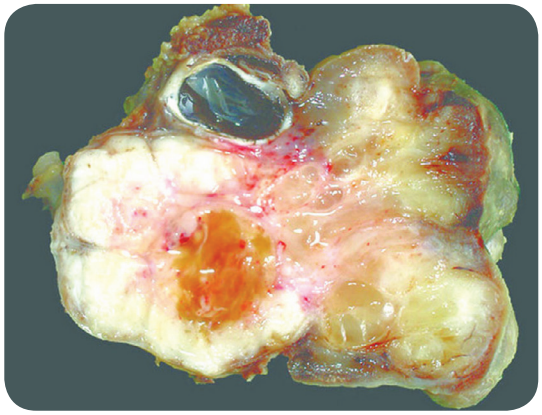

Figure 116.3 The gross pathological appearance of a rahbdomyosarcoma, embryonal subtype. The tumor is poorly circumscribed with a firm texture and is cream to white in color.

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)

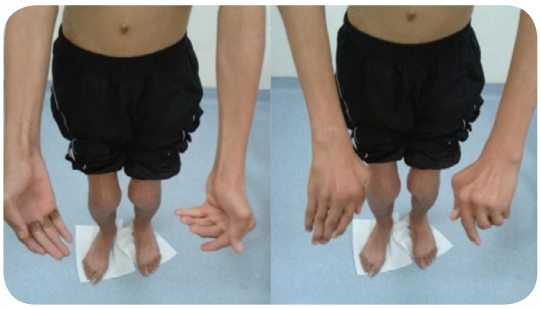

Figure 117.1 Contractures in the hands of an individual with arthrogryposis.

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

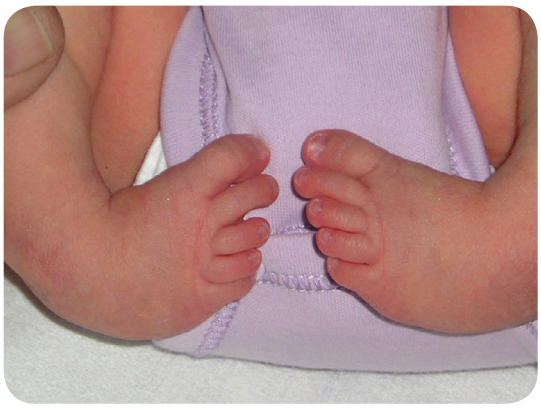

Figure 117.2 A neonate with club feet.

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

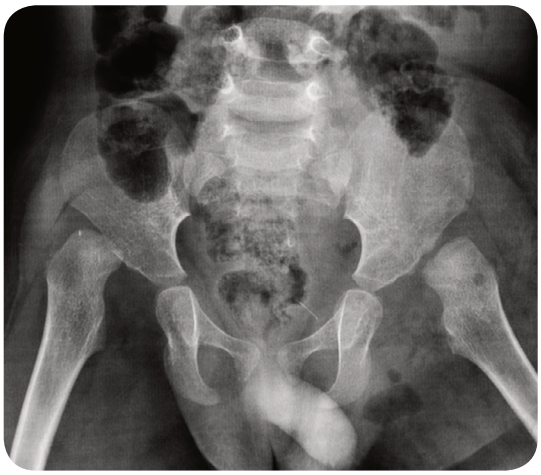

Figure 117.3 A plain radiograph of the pelvis of an infant with severe congenital hip dysplasia. There is complete dysplasia of both acetabula and superior dislocation of the femoral heads.

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

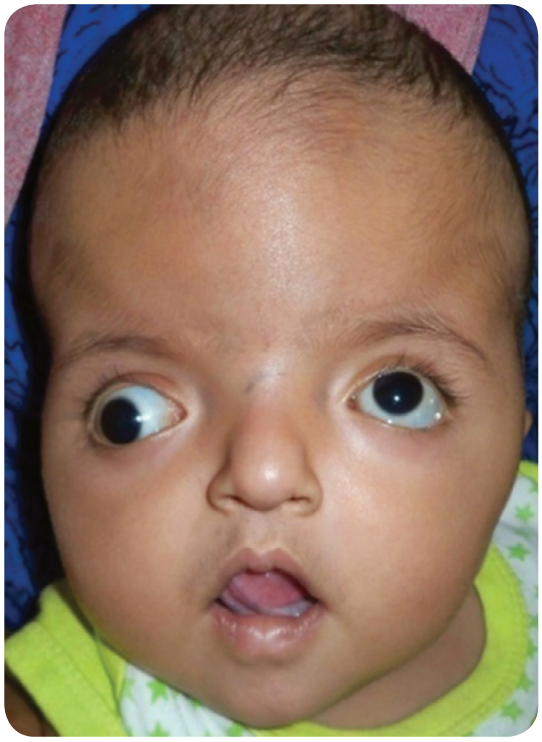

Figure 117.4 Facial features of an child with craniosynostosis in Apert syndrome.

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

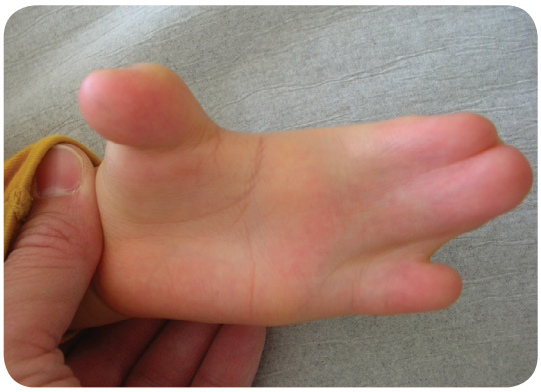

Figure 117.5 Syndactyly seen in an individual with Apert syndrome, which also causes craniosynostosis.

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

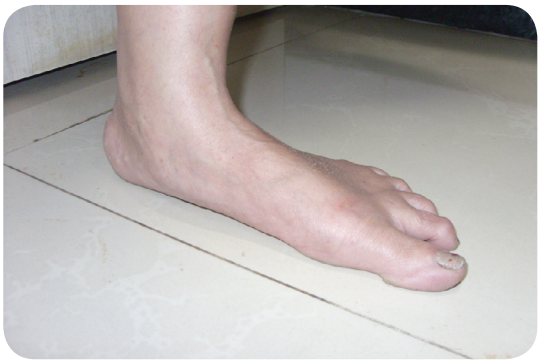

Figure 117.6 Complete collapse of the longitudinal arch has resulted in complete contact of the sole of the foot with the ground.

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)

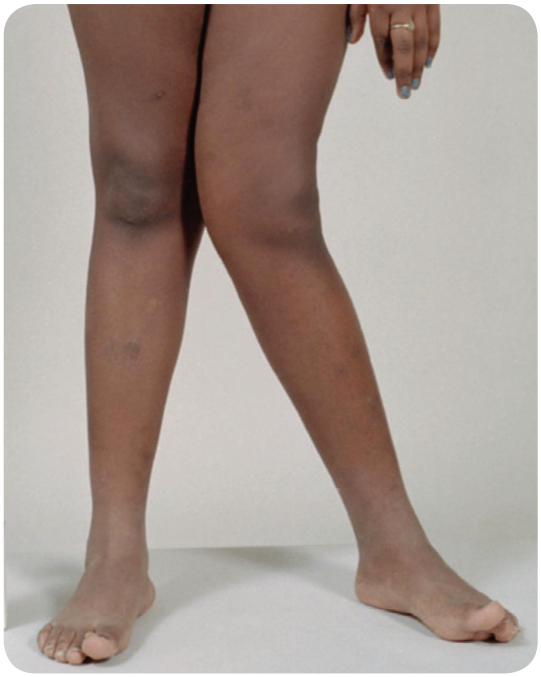

Figure 117.7 An individual with genu valgum of the left leg secondary to surgery and radiotherapy to treat a synovial sarcoma of the lateral distal femoral epiphysis as a child. The medial epiphysis continued to grow whilst growth of the lateral epiphysis was stunted.

DIAGNOSIS

- Clinical evaluation

DIAGNOSTIC IMAGING

X-ray

- Both legs (hips to feet) in standing position

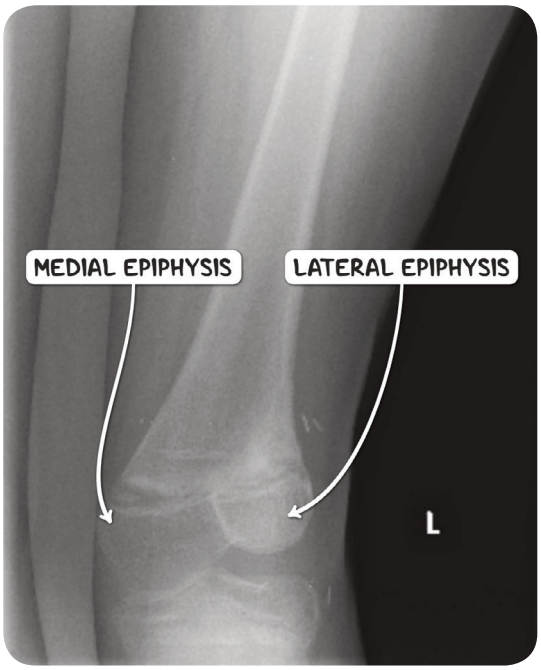

Figure 117.8 An X-ray image of the affected knee. The medial head is much larger than the lateral head.

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

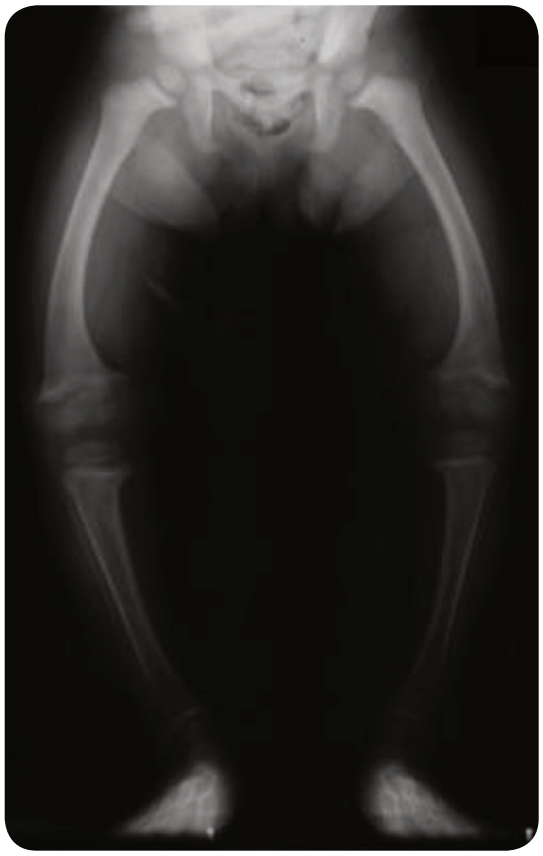

Figure 117.9 An X-ray image of a child with rickets displaying genu varum.

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

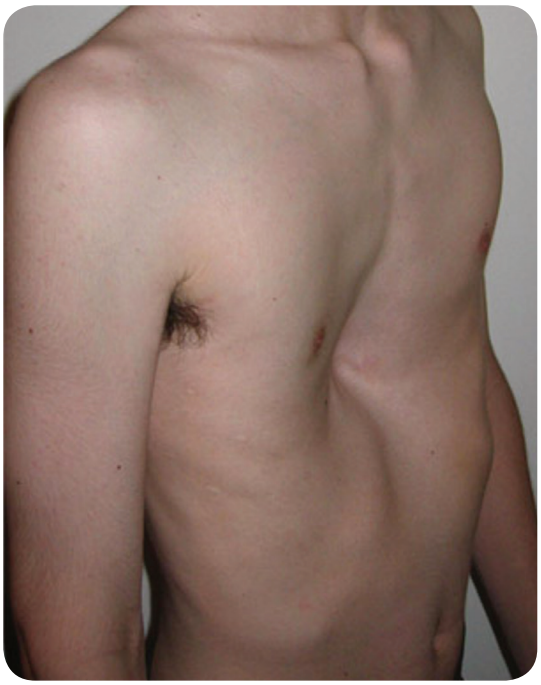

Figure 117.10 An individual with pectus excavatum.

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

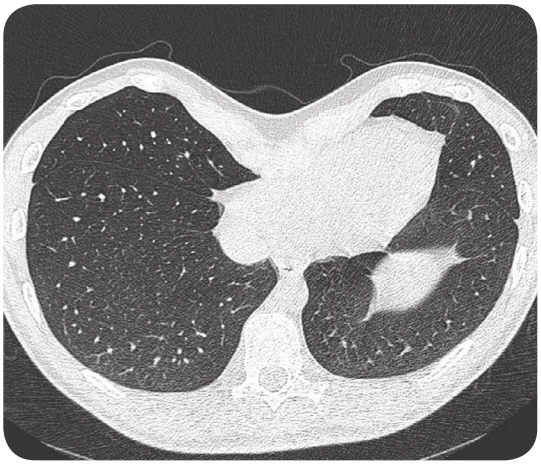

Figure 117.11 A CT scan of the chest in the axial plane demonstrating pectus excavatum.

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

NOTES
SKELETAL DYSPLASIA

GENERALLY, WHAT IS IT?

PATHOLOGY & CAUSES

- Disorders affecting bone development

CAUSES

- Inherited/sporadic (*de novo*) genetic mutation

COMPLICATIONS

- Caused by bone malformations, depend upon affected bones

SIGNS & SYMPTOMS

- Commonly involve growth impairment, bone malformation

DIAGNOSIS

DIAGNOSTIC IMAGING

X-ray/CT scan/MRI

- Bone malformation, impaired ossification visualization
- Bone-age estimation

Ultrasound

- Prenatal diagnosis

TREATMENT

SURGERY

- Bone malformation correction if warranted/ desired

ACHONDROPLASIA

osms.it/achondroplasia

PATHOLOGY & CAUSES

- Genetic disorder, causes dwarfism with disproportionate short stature
- Relatively normal-sized torso, short limbs, normal–large head (macrocephaly) with prominent forehead (hence "disproportionate dwarfism")
- Average height
  - *Biologically-male:* 131cm/4'4''
  - *Biologically-female:* 123 cm/4'

CAUSES

- Heterozygous "gain-of-function" mutation in *fibroblast growth factor receptor 3* (*FGFR3*) gene on chromosome 4; individuals with homozygous mutation usually do not survive
  - Mutated receptor displays "constitutive activity" (activated in ligand binding absence) → inhibits chondrocyte proliferation
  - Endochondral ossification affected → inhibits long bone elongation (e.g. humerus, femur)

- Intramembranous ossification less affected → relatively normal flat bone growth (e.g. skull, ribs)
- Autosomal dominant inheritance pattern (20%); sporadic mutation in most cases (80%)

COMPLICATIONS

- Eustachian tube narrowing → recurrent middle ear infection
- Narrowing of
  - Lumbar spinal canal → spinal stenosis
  - Foramen magnum → cervical medullary compression
- Venous obstruction at sigmoid sinus → hydrocephalus
- Midface retrusion → obstructive sleep apnea
- Obesity

SIGNS & SYMPTOMS

- Long bone malformations
  - Rhizomelic (proximal) limb shortening
  - Varus/valgus leg malformations
  - Short metacarpals
  - Short phalanges (brachydactyly)
  - Trident hand (fingertips cannot touch)
- Flat bone malformations (less common)
  - Enlarged head
  - Frontal bossing (prominent forehead)
  - Flattened nasal bridge (saddle nose malformation)
  - Narrow foramen magnum
  - Spinal kyphosis/lordosis

DIAGNOSIS

DIAGNOSTIC IMAGING

Prenatal ultrasound

- Skull width to femur length ratio higher than normal

X-ray/MRI/CT scan

- Large skull with small skull base
- Narrow foramen magnum, spinal canal
- Short, flattened vertebral bodies
- Small flat squared iliac wings ("mickey mouse ear")
- Fibular overgrowth
- Metaphyseal flaring: diaphysis narrowing, metaphysis widening

LAB RESULTS

- DNA test
  - *FGFR3* mutation-positive

TREATMENT

SURGERY

- Bone malformation correction warranted/desired

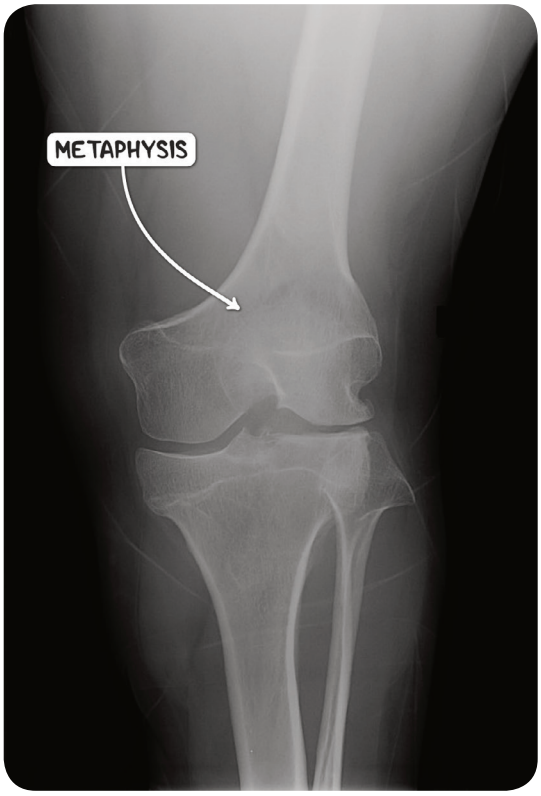

Figure 118.1 An X-ray image of the knee of an individual with achondroplasia. There is flaring of the distal femoral metaphysis typical of the disease.

CLEIDOCRANIAL DYSPLASIA

osms.it/cleidocranial-dysplasia

PATHOLOGY & CAUSES

- AKA cleidocranial dysostosis
- Skeletal dysplasia
  - Predominantly affects intramembranous ossification-derived bone development
  - Clavicles (cleido-), skull bones (-cranial)
- Abnormal teeth development, delayed cranial fontanelle closure, clavicle underdevelopment/absence, distinctive craniofacial features

CAUSES

- Heterozygous runt-related transcription factor 2 (*RUNX2*) gene mutation (transcription factor involved in osteoblast, chondrocytes differentiation → delayed ossification; 30% of cases are idiopathic)
- Autosomal dominant inheritance pattern, can be sporadic

COMPLICATIONS

- Osteoporosis (adults)

SIGNS & SYMPTOMS

- Dental abnormalities
  - Supernumerary teeth (up to 13), delayed/failed permanent teeth eruption, abnormal deciduous dentition
- Delayed cranial fontanelle closure → soft skull areas
- Short stature
- Hypoplastic/aplastic clavicles → shoulder hypermobility
- Maxilla, mandibular prognathism hypoplasia
- Midface hypoplasia, flattened nasal bridge
- Frontal/parietal bossing
- Abnormal ear ossicles → hearing loss

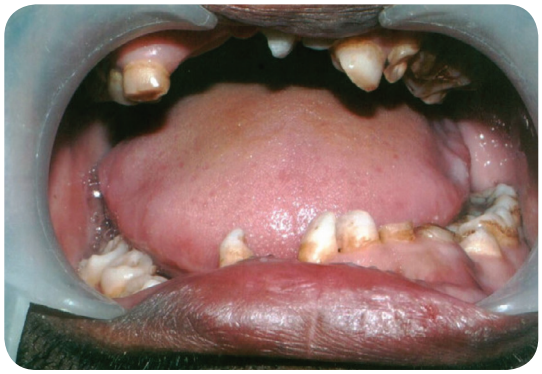

Figure 118.2 Retained deciduous teeth in the mouth of an individual with cleidocranial dysplasia.

DIAGNOSIS

DIAGNOSTIC IMAGING

X-ray

- Hypoplastic/aplastic clavicles
- Widened fontanelles
- Wormian bones (multiple small bones between sutures)
- Frontal/parietal bossing
- Supernumerary teeth
- Supernumerary ribs
- Iliac bone hypoplasia
- Symphysis pubis widening (diastasis)
- Small, highly positioned scapulas

LAB RESULTS

- Genetic testing (confirmation)

TREATMENT

SURGERY

- Bone malformation correction if warranted/desired

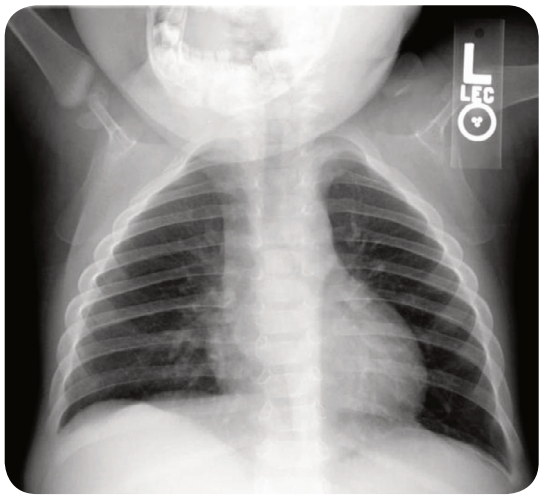

Figure 118.3 A chest radiograph of a neonate with absent clavicles, consistent with a diagnosis of cleidocranial dysplasia.

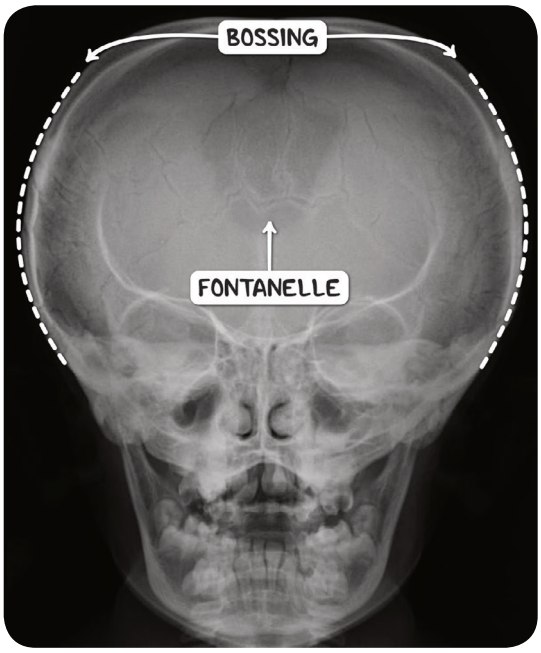

Figure 118.4 An X-ray image of the skull of a child with cleidocranial dysplasia. There is bitemporal bossing and a widened frontal fontanelle. The posterior lambdoid suture (not visible) contains multiple wormian bones.