MUSCULOSKELETAL SYSTEM DISORDERS

ALTERATIONS OF SKELETAL SYSTEM

ACHONDROPLASIA

1. Define: Impaired cartilage proliferation in the growth plate
2. Etiology:
    a. Autosomal dominant
3. Common cause of dwarfism
4. Patho: Due to activating mutation in fibroblast growth factor receptor 3 (FGFR3) leads to
5. Key features:

OSTEOGENESIS IMPERFECTA (OI)

1. Define: Congenital defect of bone formation resulting in structurally weak ones
2. Etiology: Autosomal dominant defect in collagen type 1 synthesis
3. Brittle bone dz
    a. can be classified by severity, genetic, radiographic, and clinical characteristics
4. Key characteristics:
    a. weak bone, blue sclera, hearing loss

OSTEOMALACIA (RICKETS)

1. Define: Defective mineralization of osteoid
    a. deficient mineralization at the growth plate of long bones, resulting in growth retardation.
    b. If the underlying condition is not treated, bone deformity occurs, typically causing bowed legs and thickening of the ends of long bones
2. Called Rickets <1 yr. old children
3. Key factors
    a. Vitamin D and calcium deficiency
    b. Lack of sunlight
    c. Family hx
    d. Malabsorption syndromes
4. Patho: Due to low vit d >> low calcium and phosphate >> impact the osteoblasts to produce osteoid which then decreases bone formation

PAGET DISEASE

1. Define: Chronic localized bone remodeling disorder characterized by increased bone resorption, bone formation, and remodeling, which may lead to major long bone and skull deformities
    a. Imbalance between osteoblast and osteoclast function
    b. Thick sclerotic bone that fractures easily
2. Patho: metabolic hyperactivity of the bone>> osteoclasts responsible for increased bone resorption causing large pores and pits >> high turnover reveals irregular and “woven” boney

nature >> less resistant and more elastic >>prone to microfractures especially in weight baring bones

3. Key factors
    a. Asymptomatic
    b. FMhx
    c. Bone pain- back or long bone
    d. Boney deformities

OSTEOPOROSIS

1. Define: In US 10 mill people have systemic skeletal disease
    a. low bone mass; abnormal bone architecture; compromised bone strength
    b. increase in bone fragility and risk of fracture
        i. Reduction of trabecular bone mass
        ii. Porous bone w/ increased risk of fracture
2. Based on peak bone mass and rate of bone loss
    a. What age?_____ What is the rate of loss? _____
3. Senile and post-menopausal most common
4. Risk factors:
    a. Female, white, older age, postmenopausal
    b. ETOH, smoking
    c. Nutritional
        i. Low vit D, calcium
    d. Low BMI
    e. Medications
        i. Corticosteroids
        ii. Other_____
    f. Endocrine
        i. DM, hypogonadism, amenorrhea
    g. Key Characteristics
        i. Back pain
        ii. Incidental finding on xrays
        iii. Kyphosis
        iv. Impaired gait, lower extremity weakness
        v. Vertebral tenderness
    h. FRAX scores
    i. Diagnosing Osteoporosis
        i. DXA scan
            1. T-scores: determined by measuring bone density at the hip, so are best for predicting hip fracture.
                a. Accuracy of DXA at the hip is >90%.
                b. Extrapolation of T-scores to other sites may not be as predictive of fracture.

c. T-score -/< -2.5 indicates osteoporosis
d. T-score -/< -2.5 w/ fragility fx's indicates severe or established osteoporosis

2. X-ray: Not diagnostic of osteoporosis
    a. X-ray may reveal osteopenia and/or fractures (vertebral fractures)
    b. used to drive the need for DXA assessment when osteopenia is detected coincidentally.
    c. should be considered in patients with:
        i. pain in the thoracolumbar spine
        ii. height loss
        iii. thoracic kyphosis

OSTEOMYELITIS

1. Define: Infection of bone and marrow
2. Transient bacteremia (children) seeds metaphysis
    a. Children 1 to 4 years: S.aureus , Strep pyogenes, H. influenzae
3. Open wound bacteremia (adults) seed epiphysis
    a. Age 21 + : S. aureus
4. Acute or chronic
5. Biofilm forming bacteria
6. Patho:
7. Key Characteristics:

AVASCULAR NECROSIS (OSTEONECROSIS)

1. Define: Ischemic necrosis of bone and bone marrow
2. Compromise of the bone vasculature, leading to the death of bone and marrow cells
    a. bone marrow infarction
    b. ultimate mechanical failure
3. Etiology: Trauma, fracture, steroids, sickle cell, caisson disease
    a. direct damage to bone vasculature (femoral neck fracture)
    b. direct injury of bone or marrow elements
4. Patho:
5. Key Characteristics:

DEVELOPMENTAL DYSPLASIA OF THE HIP

1. Define: Continuum of conditions affecting the proximal femur and acetabulum
    a. ranging from acetabular immaturity to hip subluxation and frank hip dislocation
2. Epidemiology: 1.5-20/1000 births /more common in females 4 xfold than males
3. DDX: rule out transient dysplasia

    a. which represents acetabular immaturity in which the anatomic relationship stabilizes
    b. normalizes over a period of weeks to months
4. Key factors
    a. Genetic, hormonal, and mechanical factors may contribute
    b. Female, breech presentation, FamHX
    c. Screening exam may reveal restricted abduction, abnormal position reported by parents
        i. Hip is grossly unstable
        ii. Barlow and Ortolani tests can help distinguish
        iii. U/S and hip Xray

CLUB FOOT (EQUINOVARUS FOOT DEFORMITY)

1. Define: Congenital talipes equinovarus
2. Patho:
3. Epidemiology:
    a. Very common birth defect accounting for 1/1000 births
    b. M>F
4. Etiology unknown/multifactorial
    a. Idiopathic, postural, or non-idiopathic classifications
    b. influenced by both environmental and genetic factors
5. Key features
    a. hindfoot in varus
    b. forefoot is adducted
    c. ankle in equines
6. Diagnostics:
    a. Ap radiographs feet, pelvis, hip U/S
    b. CT foot/spine
    c. Abd U/S

TRAUMA

FRACTURE CLASSIFICATION:

1. Complete / incomplete
2. Open(compound)
3. closed (simple)
4. comminuted

DIRECTION OF FRACTURE LINE:

1. linear, oblique, spiral, transverse, greenstick, torsos, bowing, fragility fx's
2. Pathological, stress or trans chondral

EVALUATION AND TREATMENT

1. Realigning bone fragments (reduction)
    a. WHY?

2. Hold those bone fragments in realigned position (immobilization)
    a. WHY?
3. Malunion, nonunion, or delayed union
    a. Differences between? What does each mean?
4. Dislocation and subluxation

ALTERATIONS OF JOINT SYSTEM

OSTEOARTHRITIS

1. Define: AKA Degenerative joint disease
    a. Progressive degeneration of articular cartilage
    b. "wear and tear"
2. Patho: failure to maintain homeostatic balance of if the cartilage matrix synthesis and degradation bone remodeling, bone marrow lesions, synovial inflammation cartilage loss
3. Types of osteoarthritis
    a. Primary (idiopathic): no preceding injury to the joint
        i. localized OA
            1. affects the hands, knee, hip, or foot (especially the first metatarsophalangeal)
        ii. generalized OA
            1. affects hands and another joint
    b. Secondary: an antecedent insult to the joint
        i. congenital abnormality
        ii. congenital hip dysplasia
        iii. Trauma
        iv. inflammatory arthropathies
            1. rheumatoid arthritis, chronic gout
        v. ongoing strenuous physical activities or occupations
            1. lead to joint damage over time

RHEUMATOID ARTHRITIS

1. Define: Chronic systemic autoimmune disease
    a. hallmark synovitis leading to formation of pannus (inflamed granulation tissue)
2. Epidemiology:
3. Etiology: unknown, possible genetic and infectious causes have been postulated
    a. Classically presents in women of late childbearing age >> why?
    b. Genetics:
    c. Infectious:
4. Patho:

5. Key Characteristics:

a. SYMMETRIC involvement PIP joints of the fingers, wrists, elbows, ankles, and knees are characteristic
b. Joint space narrowing, loss of cartilage and osteopenia

JUVENILE IDIOPATHIC ARTHRITIS

1. Define: A collection of chronic pediatric arthropathies/rheumatological disease in children
    a. most common chronic arthropathy of children
    b. includes several subtypes
        i. oligoarticular
        ii. Polyarticular
        iii. systemic onset (known as _____)
    c. onset before 16 years of age
2. Epidemiology:
    a. Affects 1 in 1000 children and can present at any age
3. Key Characteristics:
    a. presence of objective arthritis (in one or more joints) for at least 6 weeks
    b. Arthritis of joints is defined by swelling or effusion, increased warmth, and/or painful limited movement with or without tenderness

GOUT

1. Define: Deposition of monosodium urate crystals in the tissues, especially in joints
2. Patho: Hyperuricemia r/t overproduction or decreased excretion of uric acid
    a. causing attacks of acute inflammatory arthritis
    b. tophi around the joints and possible joint destruction, renal glomerular, tubular, and interstitial disease; and uric acid urolithiasis.
3. Key Characteristics:
    a. most commonly affects the first toe (podagra), foot, ankle, knee, fingers, wrist, and elbow; however, it can affect any joint.
    b. Swelling, effusion, warmth, erythema and/or tenderness of the involved joint(s)
4. Etiology:
5. Risk factors:

ANKYLOSING SPONDYLOSIS

1. Define: Chronic inflammatory back pain is the hallmark clinical feature
    a. back pain that has an insidious onset, is worse in the morning, and improves with exercise
    b. mainly affects the axial skeleton, although peripheral joints, entheses
        i. tendon or ligament attachments to bone
    c. extra-articular sites such as the eye and bowel are frequently affected
2. Etiology:

3. Diagnosis: 4 out of 5 must be present:
    a. Age <40 years
    b. Back pain >3 months
    c. Insidious onset
    d. Improvement with exercise
    e. Early morning stiffness.

INFECTIOUS ARTHRITIS

1. Define: Arthritis d/t infectious agent
    a. Older Children and adults =S aureus
    b. Young adults = N gonorrhea
2. Single joint, usually knee
3. Etiology:
4. Patho:

BURSITIS

1. Define: bursa is fluid filled sac adjacent to tendon/bone to reduce friction
    a. Trauma and repeated stress can cause Inflammation of bursal sacs (bursitis)
    b. Mechanisms: hemorrhage, microtrauma, inflammatory, septic
    c. Common locations:
        i.
    d. Diagnostics:
        i. Xray r/o_____
        ii. U/S
        iii. Inflammatory markers _____

SPRAIN AND STRAINS

2. Define: Tendon and ligament trauma d/y mechanical stretching, injury, trauma
    a. Tear in tendon is strain
        i. Common locations:
    b. Tear in ligament is sprain
        i. Common locations:

*****Ligament/tendon torn >>> granulation tissue containing macrophages, fibroblasts, and capillary buds surround and invade tissue >> begin repair process >> 3-4 days after injury collagen formation begins >> disorganized at first then interweave with existing collagen fibers

- 4-5 weeks after injury >>> ligament or tendon cannot withstand a strong pull- a re-injury may still occur

TENDONITIS/TENDINOSIS

3. Trauma and repeated stress can cause degradation of collagen fibers (tendinosis)

4. Inflammation of tendons (tendonitis)
5. MTU: muscle-tendon units
    a. common sites =the rotator cuff (supraspinatus tendon) in the shoulder
    b. wrist extensors (lateral epicondyle) and pronators (medial epicondylitis) in the elbow
    c. patellar and quadriceps tendon in the knee, and Achilles tendon in the heel

ALTERATIONS IN MUSCLE SYSTEM

RHABDOMYOLYSIS

1. Result of any disease process that causes muscle cell (myocyte) lysis
    a. Many etiologies:
    b. May have an obvious presentation, such as traumatic "crush" injury
    c. May be insidious, requiring a high clinical index of suspicion
2. Key Characteristics:
    a. Muscular pain or discomfort is common
    b. can have no symptoms or physical signs
    c. Crush injuries, “downtime”, immobility, and compartment syndrome
    d. Classic triad of muscle pain weakness and dk urine
3. Diagnosis
    a. elevated serum CK level
4. Patho: myoglobinuria from rapid breakdown of muscle that releases intracellular contents including protein pigment (myoglobin) into extracellular space and bloodstream.

MUSCLE STRAINS

1. Define: Mild injury usually seen after sports activity
    a. Local damage often results from sudden forced motion beyond normal capacity
2. Late complication is myositis ossificans
    a. Epidemiology:
    b. Abnormal bone formation in soft tissue
    c. Patho: is unknown but thought to be a differential issue when mesenchymal cells into osteoblasts and improper development of fibroblasts

CONTRACTURES

1. Pathological or physiological
    a. Physiological occurs with absence of muscle action in the sarcomeres
        i. muscle shortening on basis of calcium pump failure
    b. Pathological is permanent muscle shortening caused by spasm or weakness
        i. plenty of ATP and occur in despite of normal action potential

STRESS INDUCED MUSCLE TENSION

1. Define: Abnormally increased muscle tension associated with anxiety
2. Patho: abnormalities in CNS, reticular activating system, and ANS have been implicated
3. Key features
    a. Manifest as chronic pain, headaches, and neck stiffness
4. treatment would focus on progressive relaxation training, yoga, meditation, and biofeedback

FIBROMYALGIA

1. Define: Chronic musculoskeletal syndrome w/ widespread muscle & joint pain fatigue, headaches, and IBS
    a. Increased sensitivity to heat, cold, electrical stimuli
2. Epidemiology: Overall prevalence is 2% (US)
3. Patho: No pathophys process, based on subjective symptoms but altered circadian rhythms, ANS dysfunction and endocrine axis have been reported
4. Key factors
    a. 80-90% are female
    b. peak age 30-50
    c. ACR no longer uses tender point classification
        i. Use SSI (sx severity score) and WPI (widespread pain index)
5. Differential includes
    a. Chronic fatigue syndrome
    b. Vit D def
    c. RA, SLE

CHRONIC FATIGUE SYNDROME

1. Define: otherwise known as myalgic encephalomyelitis; a debilitating and complex disorder
2. diagnosis of exclusion
3. Patho: believed to be less of a muscular skeletal disorder and more related to hypersensitivity of the central nervous system
    a. also known as central sensitization exact
4. etiology and pathophysiology remain unknown
5. Key features
    a. profound fatigue
    b. musculoskeletal pain
    c. cognitive impairment
    d. unrefreshing sleep
    e. impaired neurologic energy production
    f. immune impairment
    g. individuals may present with a sore throat headache and tender lymph nodes they occur without a clear pathophysiologic explanation

DISUSE ATROPHY

1. Define: Pathological reduction in normal muscle fiber size
    a. from an accelerated degradation of it may be associated with normal aging (sarcopenia)
2. Etiologies include:
    a. prolonged inactivity from bedrest, casting, or local nerve damage
    b. prolonged immobilization and low mechanical load such as inactivity during bed rest trauma (via casting) local nerve damage or spaceflight
    c. disease induced atrophy
        i. seen in debilitating conditions such as cancer, aids, renal failure, congestive heart failure, COPD and burns

MCARDLE DISEASE

1. Define:
2. Etiology: Autosomal recessive disease
    a. 1st myopathy with a single enzyme defect identified
    b. Glycogen myophosphorylase deficiency
        i. Responsible for glycogen breakdown – muscles rely on CHO and fatty acids for energy , defect in this gene means person unable to break down glycogen or lactate for muscle energy
3. Key Characterizations:
    a. Manifests itself as exercise intolerance, fatigue, and painful muscle cramps
    b. if exercise is carried to an extreme painful muscle contracture and myoglobinuria developed as the disease progresses some individuals have pronounced weak muscles and wasting

INFLAMMATORY MUSCLE DISEASES

MYOSITIS

1. Define: Induced inflammatory changes in skeletal muscle
2. Etiologies include:
    a. viral, bacterial, and parasitic infections w/ varying severity induce inflammatory changes within the muscle
        i. TB, sarcoidosis, trichinellosis
3. self-limiting symptoms of muscle aches and pains during influenza may be in acute subacute form of viral myopathy
4. Key characteristics:
    a. Muscle pain, tenderness, signs of inflammation and elevation of creatinine kinase
    b. common manifestations of viral myositis
    c. tuberculosis and sarcoidosis chronic inflammatory changes in granulomas are found in the muscle

NEUROMUSCULAR DISORDERS

MUSCULAR DYSTROPHY

1. Define: Degenerative disorder characterized by muscle wasting and replacement of skeletal muscle by adipose tissue
    a. progressive, generalized diseases of muscle, most often caused by defective or specifically absent glycoproteins (dystrophin) in the membrane of the muscle wall
    b. All muscular dystrophies are characterized by ongoing degeneration and regeneration of muscle fibers
2. The most common and rapidly progressive muscular dystrophy is Duchenne muscular dystrophy (DMD)
    a. This is X-linked and diagnosed by the finding of absent dystrophin on muscle biopsy
    b. defects in dystrophin gene
    c. DUCHENNE d/t gene deletion
3. Other genetic
    a. Becker
    b. Emery Dreifuss

CEREBRAL PALSY

1. umbrella term referring to a non-progressive disease of the brain originating during the prenatal, neonatal, or early postnatal period when brain neuronal connections are still evolving
2. Most common cause of childhood disability affecting 2.5 per 1000 individuals in the industrialized world
3. All patients present with motor impairment
4. 80% have spasticity
5. Other movement disorders observed are dystonia, athetosis, chorea, and ataxia.

MUSCULOSKELETAL TUMORS

OSTEOSARCOMA

1. Malignant proliferation of osteoblasts
2. Arises in the distal femur and proximal tibial (metaphysis of long bones)
3. Most common non-hematological bone neoplasm
4. Epidemiology: 2nd-3rd decade, peak incidence 13-16 years.
    a. M>F
    b. Peak incidence seen in teenagers
5. Presents as pathologic FX or bone pain and swelling
6. No known etiology for primary

RHABDOMYOSARCOMA

1. Define: group of rare solid tumors of connective tissue
    a. Most common soft tissue sarcoma in childhood
    b. 2 types of soft tissue or bone
        i. 1% adult malignancy/15% pediatric malignancy
        ii. 50 subtypes are known
2. Etiologies include
    a. Unknown, infection (Kaposi) HSV8
    b. Radiation
    c. Vinyl chloride
3. Key characteristics:
    a. commonly presents as soft-tissue swelling
    b. may or may not be painful
    c. Upper/lower GI bleed
    d. Rash
    e. DUB
    f. Increased abd. girth
4. Differential diagnoses
    a. lymphoma, metastatic carcinoma
    b. benign lesions: lipoma & neuroma
5. Patho: not understood, no theories.