Pathophysiology 2

Skeletal System Conditions

Congenital Orthopedic Disorders & Diseases

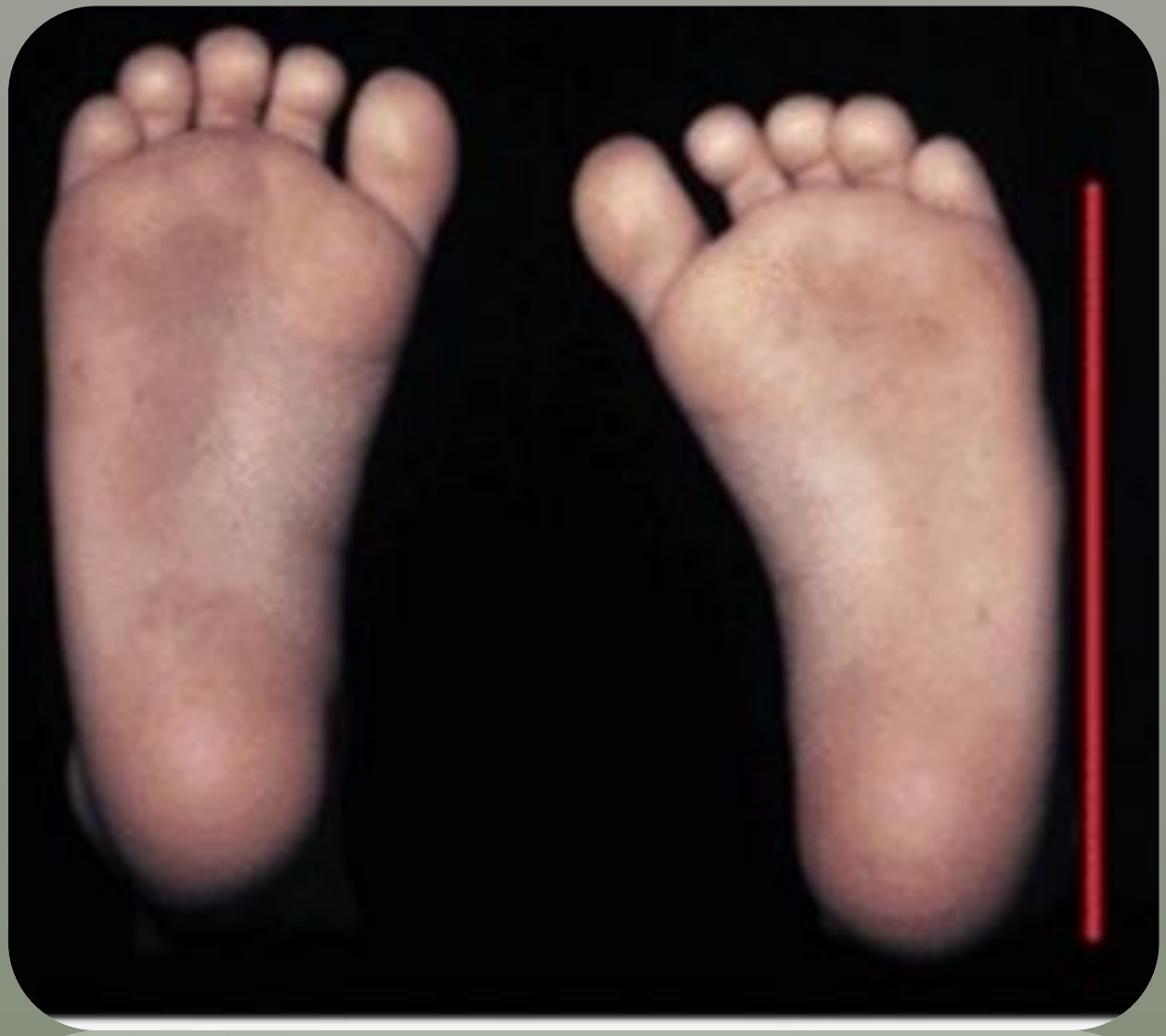
- Disorders that affect the musculoskeletal system
- AKA orthopedic birth defects
- Occur while the baby is still developing in the womb and appear at birth.
- Caused by a wide range factors
  - gene abnormalities
  - toxin ingestion
  - disease and illness during pregnancy
- different types and may be characterized by different symptoms
- may or may not require treatment

Topics Covered

- Metatarsus Adductus
- Clubfoot
- Developmental Dysplasia of the Hip (DDH)
- Osteogenesis Imperfecta
- Muscular Dystrophy
- Congenital Scoliosis
- Hypermobility Syndrome

Metatarsal Adductus

- (AKA metatarsus varus)
  - common foot deformity noted at birth that causes the forefoot, deviate medially
- 2 Types
  - "flexible" (the foot can manually be straightened to a degree)
  - "nonflexible" (the foot cannot be straightened manually).



Cause & Statistics

- Unknown etiology
- Occurs in approximately 1 to 2 per 1,000 live births
- More common in firstborn
- Increased risk for developmental dysplasia of the hip (DDH)

Diagnosis & Treatment

- Diagnosis of metatarsus adductus is made with a physical examination, complete birth history and family history of M.A.
- X-rays often done in the case of nonflexible metatarsus adductus
- An infant with metatarsus adductus has a high medial arch and the big toe has a wide separation from the second toe and deviates medially.

- Specific treatment for metatarsus adductus will be determined based on:
  - Age, overall health, and medical history
  - extent of the condition

Goals of Treatment

- Straighten the position of the forefoot and heel
- Treatment options vary for infants and may include:
  - Observation, for those with a supple, or flexible, forefoot
  - Stretching or passive manipulation exercises (joint mobilizations)
  - Casts
  - Surgery
- Studies have shown that metatarsus adductus may resolve spontaneously in the majority of affected children.

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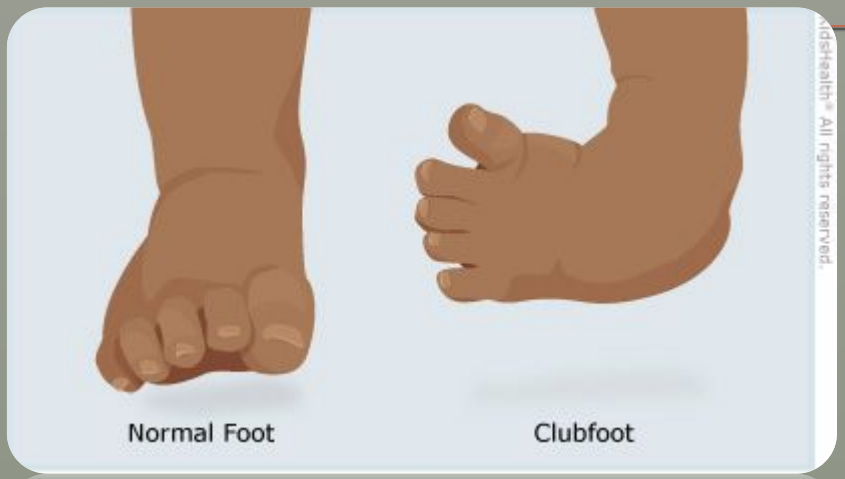
Club Foot



Club Foot

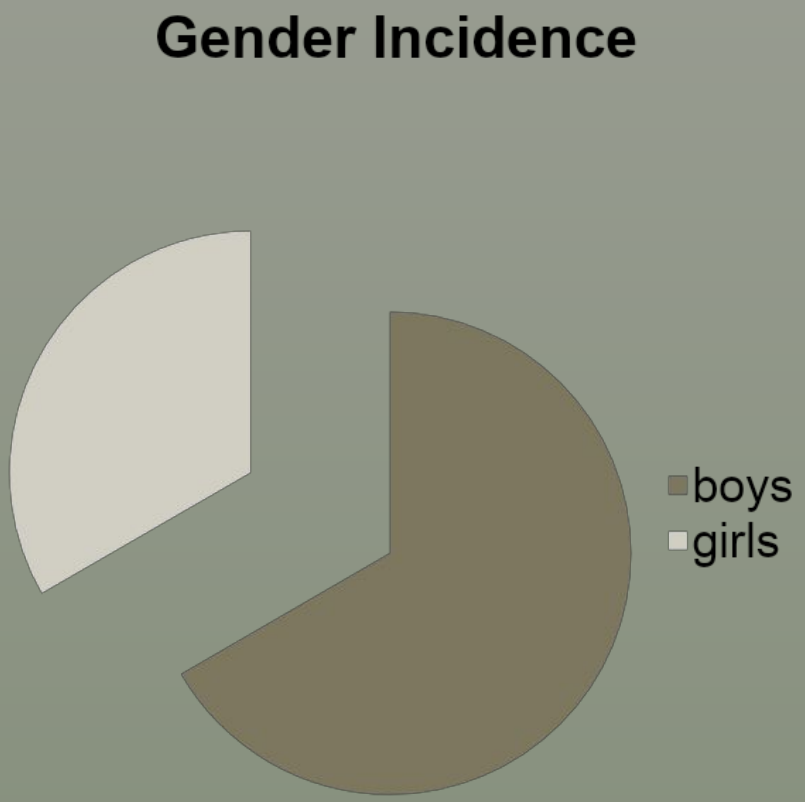
- Congenital foot deformity
- Affects bones, muscles, tendons, and blood vessels
- one or both feet
- Foot is usually short and broad in appearance and the heel points downward while the forefoot turns inward. Achilles tendon is tight
- Painless condition

Causes

- Clubfoot is considered a "multifactorial trait."
- Factors are usually both genetic and environmental. Risk factors may include:
  - Family history of clubfoot
  - Multiple gestations (twins or triplets)
  - Position of the baby in the uterus
  - Increased occurrences in those children with neuromuscular disorders.
  - decreased amount of amniotic fluid surrounding the fetus in the uterus during pregnancy - **Oligohydramnios**
- Babies born with clubfoot may also be at increased risk of DDH

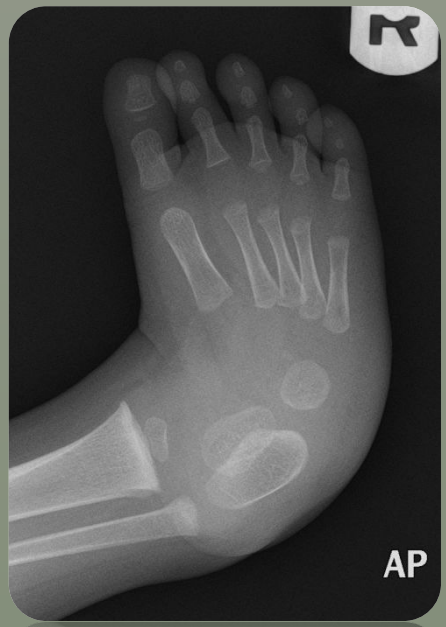
Statistics

- Clubfoot occurs in approximately 1 to 3 of every 1,000 births, with boys outnumbering girls 2 to 1
- One or both feet may be affected.



Diagnosis

- Made with a physical examination
- Doctor obtains a complete prenatal and birth history & any family history of clubfoot
- X-ray of foot may be done



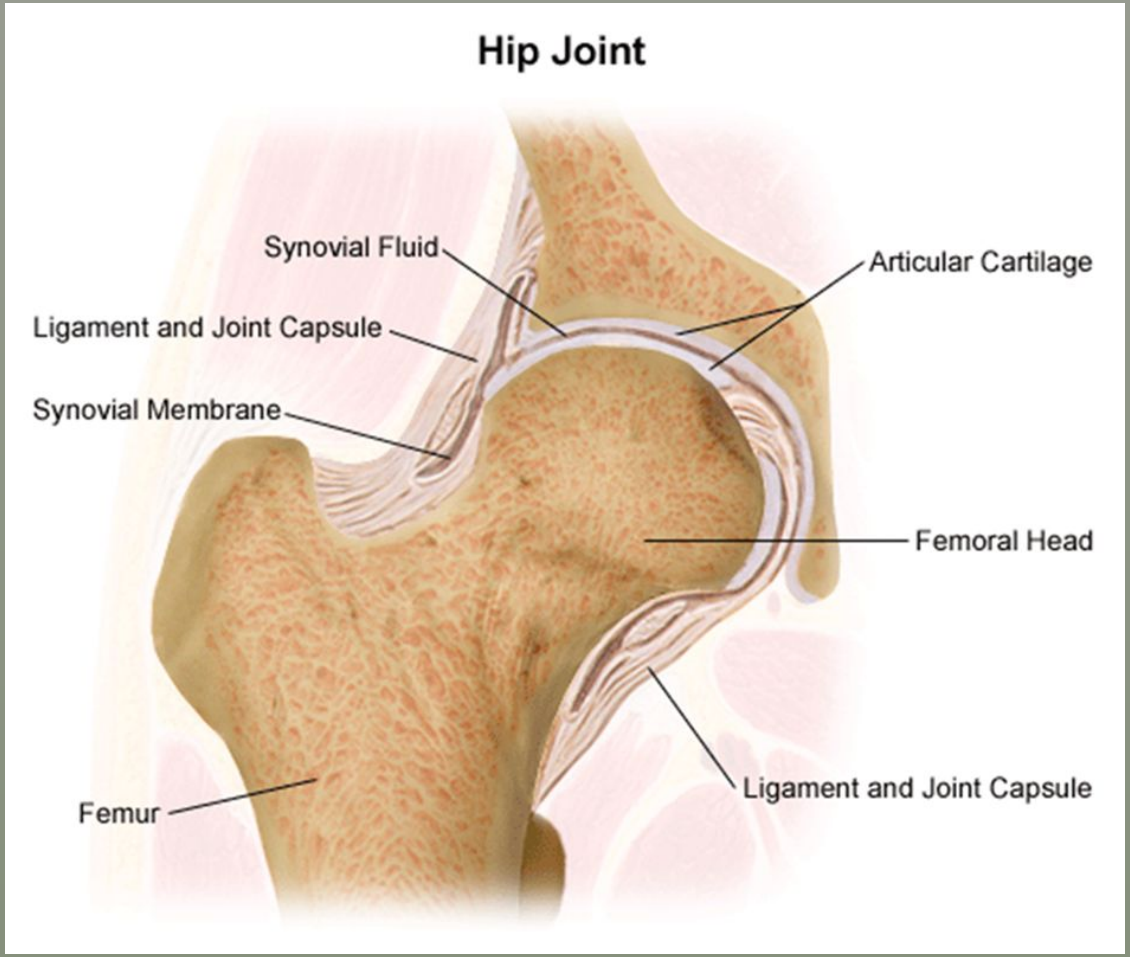
Treatment Options

- The goal of treatment is to straighten the foot so that it can grow and develop normally.
- Treatment options for infants include:
  - Nonsurgical treatment:
    - These methods include serial manipulation and casting, taping, physical therapy and splinting, and use of a machine that provides continuous passive motion.
  - A nonsurgical treatment should be the first type of treatment for clubfoot,
  - Surgery:
    - Used when nonsurgical treatment fails, or when deformity recurs and does not respond to nonsurgical treatment.

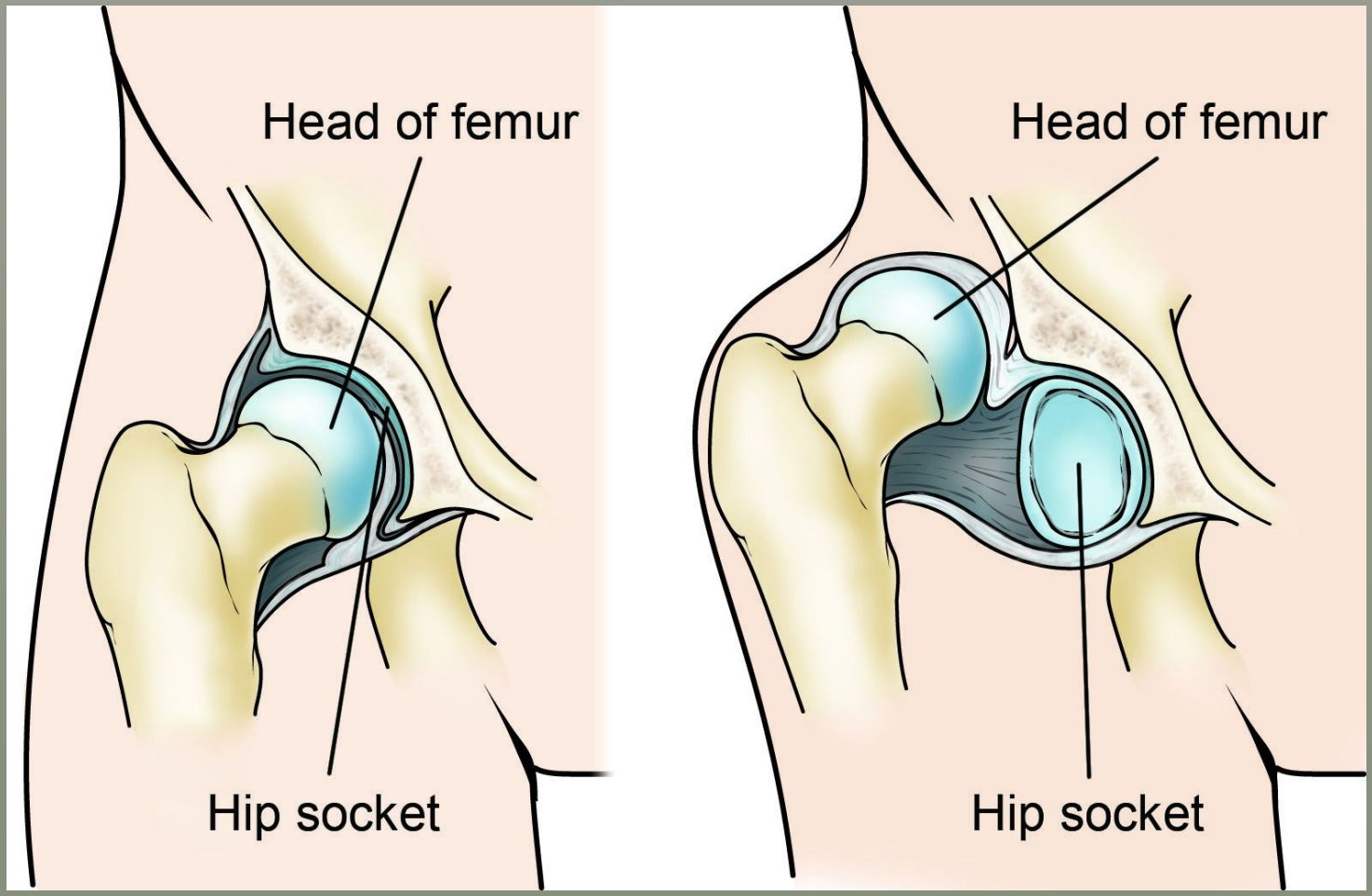
  https://youtu.be/Z4errSBsD7M

Developmental Dysplasia of the Hip (DDH)

- Congenital condition of the hip joint
- The hip socket may be shallow, letting the femoral head, slip in and out of the socket. The "ball" may move partially or completely out of the hip socket.


Hip Joint
Synovial Fluid
Articular Cartilage
Ligament and Joint Capsule
Synovial Membrane
Femoral Head
Femur
Ligament and Joint Capsule

DDH



Causes

- Considered a "multifactorial trait."
- Environmental influences thought to contribute as the baby's response to the mother's hormones during pregnancy
- Tight uterus that prevents fetal movement or a breech delivery. Left hip is involved more frequently than the right due to intrauterine positioning
- First-born babies are at higher risk
- Family history of developmental dysplasia of the hip
- very flexible ligaments

Symptoms

- Symptoms may include:
  - The leg may appear shorter on the side of the dislocated hip
  - The leg on the side of the dislocated hip may turn outward
  - The folds in the skin of the thigh or buttocks may appear uneven
  - The space between the legs may look wider than normal

Continued

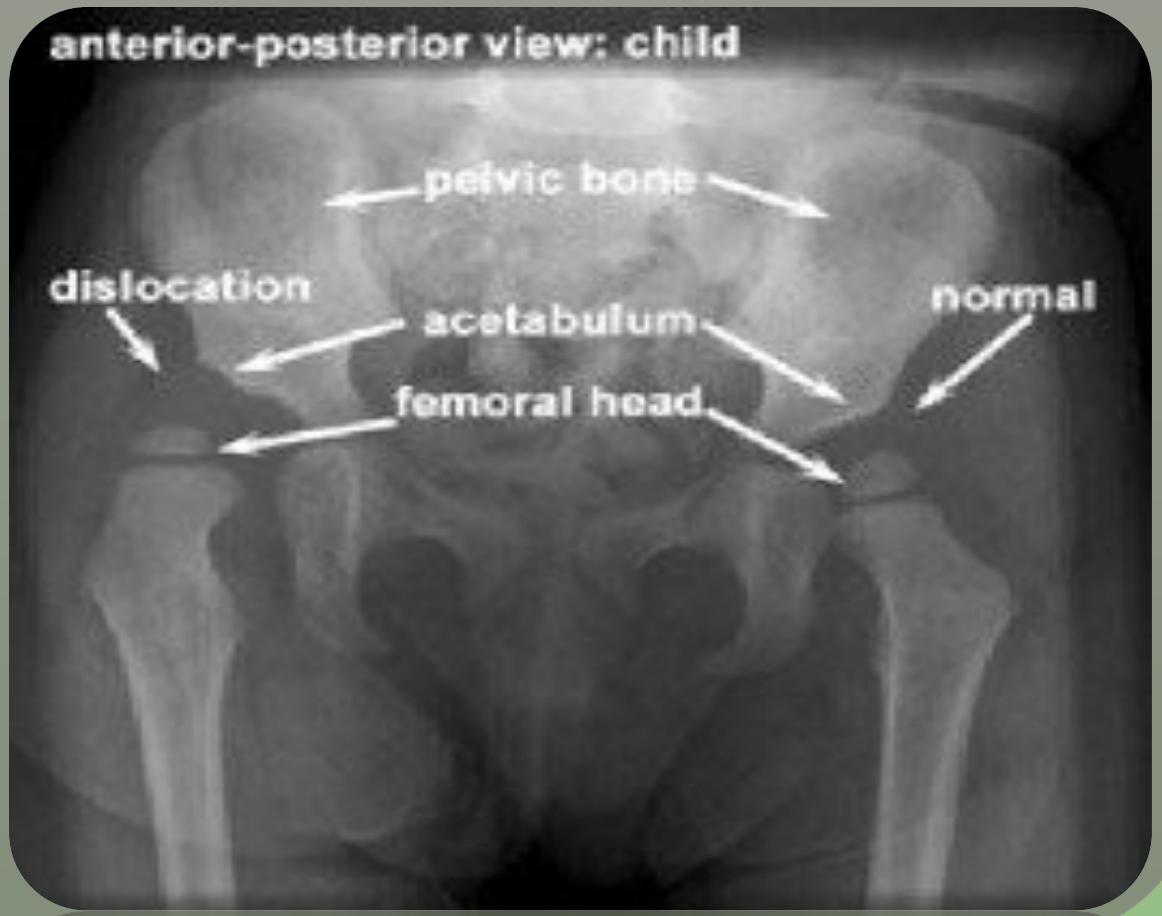
- May have a hip that is partially or completely dislocated
- Sometimes noted at birth. However, DDH may not be discovered until later evaluations.

Statistics

- The greatest incidence of DDH occurs in first-born females with a history of a close relative with the condition
- occurs once in every 1,000 live births.

Diagnoses

- Diagnostic procedures may include:
  - X-ray.
  - Ultrasound



Treatment

- The goal of treatment is to put the femoral head back into the socket of the hip so that the hip can develop normally.
- Treatment options:
  - Nonsurgical positioning **(Pavlik Harness)** is used on babies **up to 6 months of age** to hold the hip in place, while allowing the legs to move a little.
  - **Casting**- If the hip continues to be partially or completely dislocated, casting, or surgery may be required.
  - **Surgery**- If the other methods are not successful, or if DDH is diagnosed at age 6 months to 2 years, surgery may be required.

Pavlik Harness

Osteogenesis Imperfecta

- **AKA - brittle-bone disease**

- Genetic disorder characterized by bones that break easily without a specific cause.
- Can affect males and females of all races
- Believed to be because of a genetic defect that causes imperfectly formed or an inadequate amount of bone **collagen**
- To date, no known treatment, medicine, or surgery will cure
- The goal of treatment is to prevent deformities and fractures and allow the child to function as independently as possible.

Treatment

- Care of fractures
- Surgery
- Rodding, a procedure to insert a metal bar the length of a long bone to stabilize it and prevent deformity
- Dental procedures
- Physical therapy
- Assistive devices

Muscular Dystrophy

- Causes the muscles in the body to become very weak
- Muscles break down and are replaced with fatty deposits over time.
- Most common forms of muscular dystrophy are Duchenne muscular dystrophy (DMD) and Becker muscular dystrophy.
- Very similar, but Becker muscular dystrophy is less severe
- Girls are rarely affected by either of these two forms of muscular dystrophy.
- Muscular dystrophy is usually diagnosed in children between 3 and 6 years of age.

Signs & Symptoms

- Early signs
  - delay in walking, difficulty rising from a sitting or lying position, and frequent falling
  - weakness typically affecting the shoulder and pelvic muscle are one of the initial symptoms

Other S & S

- The following are the most common symptoms of muscular dystrophy
  - Difficulty climbing stairs
  - Frequent falls
  - Unable to jump or hop normally
  - toe walking
  - Leg pain
  - Facial weakness
  - Inability to close eyes or whistle

Diagnoses

- Diagnostic tests may include:
  - **Blood tests.** These include genetic blood tests.
  - **Muscle biopsy.** The primary test used to confirm diagnosis
  - **Electromyogram (EMG).** A test to check if the muscle weakness is a result of destruction of muscle tissue rather than nerve damage.
  - **Electrocardiogram (ECG or EKG).** A test that records the electrical activity of the heart, shows abnormal rhythms (arrhythmias or dysrhythmias), and detects heart muscle damage.

Treatment

- To date, there is no known treatment, medicine, or surgery
- Treatment is to prevent deformity and allow the child to function as independently as possible

Muscular Dystrophy Support

- Nonsurgical interventions may include:
  - Physical therapy
  - Positioning aids used to help the child sit, lie, or stand
  - Braces and splints used to prevent deformity, promote support, or provide protection
  - Medications
  - Nutritional counseling
  - Psychological counseling

https://youtu.be/DGOmN6rnsNk

Congenital Scoliosis

- Lateral deviation of the spinal column that may or may not include rotation or deformity of the vertebrae
- Disturbances in vertebral development during the sixth to eighth week of fetal development
- Least common form of scoliosis; it affects only 1 in every 10 000 live births

Hypermobility Syndrome

- condition that features joints that easily move beyond the normal range expected for a particular joint. Tends to be inherited.
- Often causes no symptoms and requires no treatment. Treatments are customized for each individual based on their particular manifestations.
- It is felt that certain genes (that are responsible for production of collagen) predispose to the development of hypermobile joints
- Joint hypermobility is also a feature of a rare, inherited, more significant medical condition called Ehlers-Danlos syndrome, which is characterized by weakness of the connective tissues of the body. Also commonly seen in people with Down syndrome and in people with Marfan's syndrome

Acquired Conditions

Fractures
Benign & Malignant Bone Tumours
Infectious Causes
Metabolic Bone Diseases
Arthritis
Autoimmune Conditions
Others

Fractures

- fracture is a broken bone
- may be completely fractured or partially fractured in any number of ways (crosswise, lengthwise, in multiple pieces)
- severity depends on the force
- If bone fragments stick out through the skin, or a wound penetrates called an "open" fracture
- once the skin is broken, infection in both the wound and the bone can occur

Types of Fractures

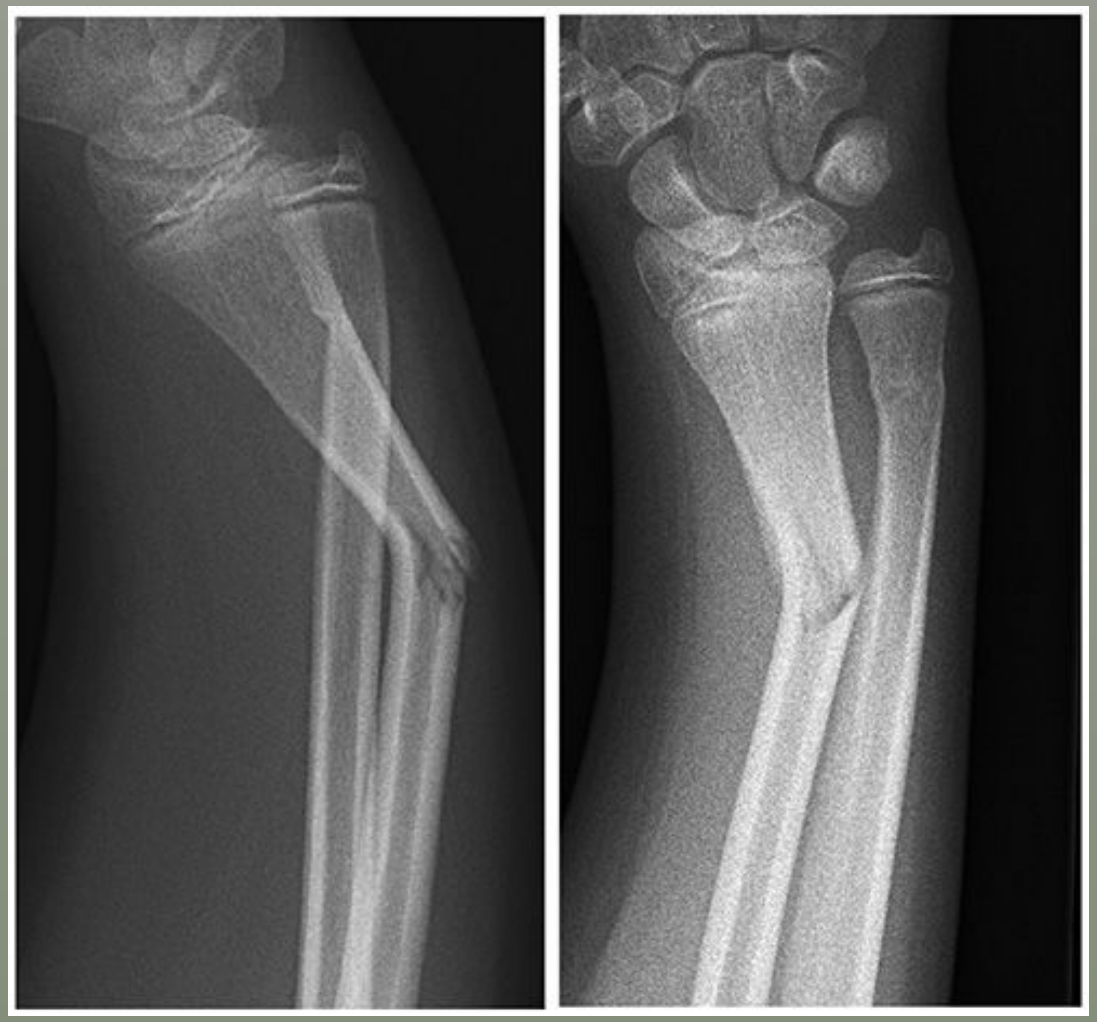
- Stable fracture
  - The broken ends of the bone line up and are barely out of place.
- Open, compound fracture
  - breaks the skin at the time of the fracture. The bone may or may not be visible in the wound
- Transverse fracture
  - horizontal fracture line
- Oblique fracture
  - angled pattern
- Comminuted fracture
  - bone shatters into three or more pieces

Pediatric fractures

- The anatomy and biomechanics of pediatric bone differ from that of adult bone, leading to unique pediatric fracture patterns, healing mechanisms, and management
- pediatric bone less dense, more porous and penetrated throughout by capillary channels
- The mechanisms of fracture change as children age.
- Younger children likely to sustain a fracture while playing and falling on an outstretched arm
- Older children tend to injure themselves while playing sports, riding bicycles, and in motor vehicle accidents.
- Child ligaments are stronger than those of an adult, forces which would tend to cause a sprain in an older individual will be transmitted to the bone and cause a fracture in a child.

Common Pediatric Fractures

- Buckle fracture
  - Compression failure of bone that usually occurs at the junction of the metaphysis and the diaphysis
  - Commonly seen in distal radius.
  - stable
  - Heal in 3-4 weeks with simple immobilization.
- Greenstick fracture
  - Bone is bent and the tensile/convex side of the bone fails.
  - Fracture line does not propagate to the concave side of the.



Complete Fractures

- Fracture completely propagates through the bone
  - spiral, transverse, or oblique, depending on the direction of the fracture line.
- Spiral fractures:
  - Created by a rotational force.
  - Low-velocity injuries
  - An intact periosteal hinge enables the orthopedic surgeon to reduce the fracture by reversing the rotational injury.
- Oblique fractures:
  - Unstable, therefore alignment is necessary.
  - Fracture reduction is attempted by immobilizing the extremity while applying traction.]
- Transverse fractures:
  - Created by a 3-point bending force.
  - Easily reduced by using the intact periosteum from the concave side of the fracture force.

Bone Tumors

- Tumors occur when cells divide abnormally and uncontrollably.
- Bone tumors form in your bones. As the tumor grows, abnormal tissue can displace healthy tissue
  - Benign Tumors
    - won't spread to other parts of the body and are unlikely to cause death, they can still be dangerous and may require treatment
  - Malignant Tumors
    - can cause cancer to spread throughout the body.

Benign Bone Tumors

- Osteochondromas
  - most common
  - Between 35 and 40% of all benign bone tumors
  - Develop in adolescents and teenagers
  - Formed of a combination of bone and cartilage
  - considered to be an abnormality of growth
  - may develop one or many

CONTINUED

- Non-Ossifying Fibroma
  - non-aggressive
  - Consists mainly of fibrous tissue
  - usually occurs in the Femur or Tibia
  - usually produces no symptoms and resolves by itself

Contined

- Giant Cell Tumors
  - Aggressive but rare
  - Grows at the ends of the body's long bones. Most often, the lower end of the femur or upper end of the tibia, close to the knee joint.
  - typically occur in young adults, and are slightly more common in females
  - can destroy the surrounding bone. Treatment for a giant cell tumor almost always involves surgery

Malignant Bone Tumors

- Primary bone cancer = originated in the bones. Primary bone cancer accounts for less than one percent of all types of cancer. The three most common forms of primary bone cancers are:
  - 1) Osteosarcoma:
  - occurs mostly in children and adolescents
  - second most common type
  - usually develops around the hip, shoulder, or knee near growth plates of bones
  - Aggressive and likely to spread to other parts of the body
  - AKA osteogenic sarcoma.
  - Terry Fox's type of cancer

Primary bone cancers Continued

- 2) Ewing sarcoma
  - Adolescents and young adults, but can sometimes affect children as young as 5 years old.
  - Usually shows up in the legs, pelvis, Spine, ribs, and upper arms and in the skull. It begins in the medullary cavities
  - African American children seldom develop Ewings sarcoma
  - More common boys than girls
  - Rapidly growing and spreading tumor.
- 3) Chondrosarcoma
  - Middle-aged people and seniors
  - Hips, shoulders, and pelvis are the usual areas for this type of bone cancer

Secondary Bone Cancers

- cancer started somewhere else in the body and then spread to the bone. It usually affects older people. The types of cancer most likely to spread to your bones are:
  - kidney
  - breast
  - prostate
  - lung
  - thyroid gland
- **Most common type of secondary bone cancer is multiple myeloma:**
  - Tumors in the bone marrow
  - People between the ages of 50 and 70 are most commonly affected

Infectious Conditions affecting Bones & Joints

Osteomyelitis
Rheumatic Fever
Septic Arthritis

Osteomyelitis

- Infection in a bone
- can reach a bone by traveling through the bloodstream ,spreading from nearby tissue or begin in the bone itself if an injury exposes the bone to germs
- In children, most commonly affects the long bones of the legs and upper arm, while adults are more likely to develop osteomyelitis in the vertebra
- Diabetics may develop osteomyelitis in their feet if they have foot ulcers
- Once considered an incurable condition, it can be successfully treated today
  - Treatment includes surgery to remove parts of the bone that have died, followed by strong antibiotics delivered intravenously for at least six weeks. Different types of bacteria typically affect the different age groups.

Signs & Symptoms

- Fever or chills
- Irritability or lethargy in young children
- Pain in the area of the infection
- Swelling, warmth and redness over the area of the infection
- Sometimes causes no signs /symptoms or has signs /symptoms that are difficult to distinguish from other problems.

Causes

- Most cases are caused by **staphylococcus** bacteria
- Germs can enter a bone in a variety of ways, including:
  - Via the bloodstream
    - Germs in other parts of your body(ex – UTI) can travel through your bloodstream to a weakened spot in a bone. In children, osteomyelitis most commonly occurs in the growth plates
  - From a nearby infection
    - Severe puncture wounds can carry germs deep inside your body. If such an injury becomes infected, the germs can spread into a nearby bone.
  - Direct contamination
    - EX – open Fracture, during surgeries

Diagnoses and Treatment

- Blood tests
- Imaging tests: X-rays, CT, MRI
- Bone biopsy

- Most common Tx are
  - Antibiotics
  - surgery to remove portions of bone that are infected or dead
  - Hospitalization is usually necessary

Rheumatic Fever

- an inflammatory disease that can develop as a **complication** of inadequately treated streptococcal throat infection or scarlet fever, caused by **group A streptococcus bacteria**
- most common in 5- to 15-year-old children
- strep throat is common, rheumatic fever is rare in developed countries. but common in many developing nations
- Can cause permanent damage to the heart, including damaged heart valves and heart failure
- Tx can reduce tissue damage from inflammation, lessen pain and other symptoms, and prevent reoccurance
- symptoms may vary. Some people may have several symptoms, others experience few. Symptoms may change during the course of the disease
- onset of rheumatic fever usually occurs about two to four weeks after a strep throat infection

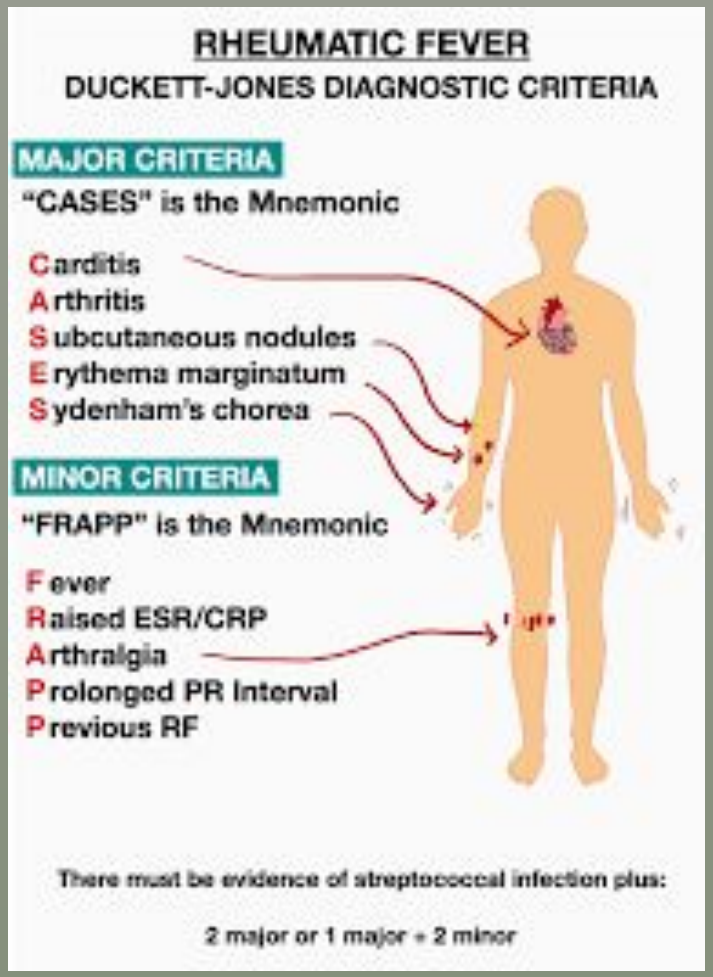
Signs and Symptoms

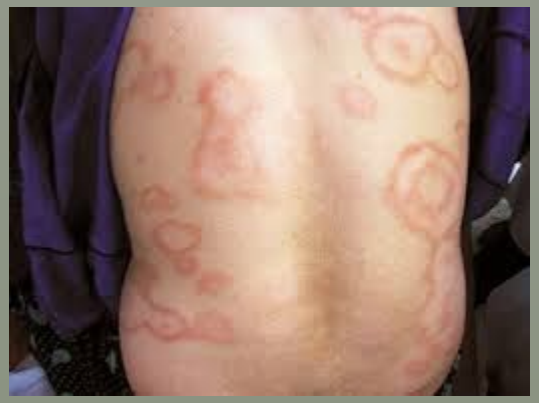
- Fever
- Painful and tender joints — most often the ankles, knees, elbows or wrists; less often the shoulders, hips, hands and feet
- Pain in one joint that migrates to another joint
- Red, hot or swollen joints
- Small, painless nodules beneath the skin
- Chest pain
- Heart murmur
- Fatigue
- Flat or slightly raised, painless rash with a ragged edge
- Jerky, uncontrollable body movements most often in the hands, feet and face

Risk Factors

- Family history – may carry a gene or genes that make them more likely to develop rheumatic fever.
- Type of strep bacteria
- Environmental factors - overcrowding, poor sanitation and other conditions that result in the rapid transmission or multiple exposures to strep bacteria.

Rheumatic Fever





Complications of Rheumatic Fever

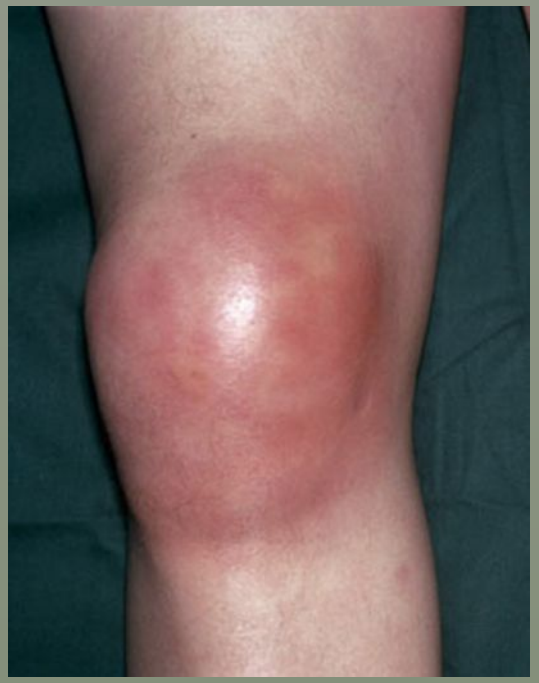
- Inflammation caused by rheumatic fever may last for a few weeks to several months. Inflammation may cause long-term complications. The damage may affect the heart muscle or valves, leading to:
  - Atrial fibrillation
    - an irregular and chaotic beating of the atria
  - Heart failure
    - inability of the heart to pump enough blood to the body

Septic Arthritis

- Intensely painful infection in a joint
- Can become infected with germs that travel through the bloodstream from another part of the body
- Also can occur when a penetrating injury brings germs directly into the joint
- More common in Infants and older adults
- Most common joints affected =knees & hips.
- Can quickly and severely damage the cartilage and bone within the joint, so prompt treatment is crucial
- Tx involves draining the joint with a needle or surgery. IV antibiotics may be necessary to stop the infection
- Typically causes extreme discomfort and difficulty using the affected joint. The joint usually swollen, red and warm, and the patient may get fever.

Causes

- Can be caused by bacterial, viral or fungal infections
- Bacterial infection with Staphylococcus aureus is most common



Metabolic Bone Disorders

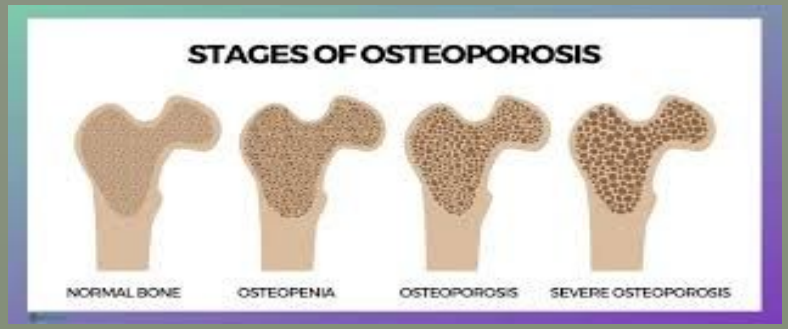
- Disorders of bone strength, usually caused by abnormalities of minerals (calcium, phosphate etc), vitamin D, bone mass or bone structure
- Can affect people of all ages
- Most common disorders include:
  - **Osteoporosis (most common of all)**: loss of bone density
  - **Osteopenia:** mild loss of bone density (pre-osteoporosis)
  - **Osteomalacia**
  - **Rickets**
  - **Paget Disease:**
  - progressive skeletal disorder that involves excessive bone destruction and repair.

Osteoporosis

Loss of normal bone density - bone becomes brittle & weak, increasing the risk of pathological (aka fragility) fractures which are fractures caused by minor force or even activities of daily life. Without treatment, it can progress to chronic pain, loss of height, stooped posture & serious disability.

How is it diagnosed?

A **Bone Density Test** is considered diagnostic. A low amount of radiation will be used to develop your **T score**, which is a comparison of your bone density compared to a healthy young adult of your sex.
A T Score of: -1 and above = normal bone density
Between -1 and -2.5 = Osteopenia (pre-osteoporosis)
Below - 2.5 = Osteoporosis



Osteoporosis

Risk Factors:

- 1) Decrease in Estrogen/Testosterone - These sex hormones help stimulate osteoblastic activity, so if they decrease then you may have more bone destruction than remodelling. Estrogen levels in women drop after menopause, putting women at greater risk
- 2) Older age
- 3) Genetic predisposition, family history, small body size
- 4) Poor lifestyle habits - Sedentary lifestyle, Smoking, Alcohol use
- 5) Medications - Long-term corticosteroids, methotrexate & others
- 6) Dietary Factors - anorexia, low intake of Calcium, Magnesium, Phosphates or Vit D, long term excessive caffeine
- 7) Diseases of Absorption - Crohns, Celiac etc

Osteopenia/Osteoporosis Treatments

1)Diet & Lifestyle changes:
Low-impact, Weight bearing exercise
Adequate consumption of bone building minerals & vitamins
Decrease/stop adverse habits (smoking etc)

2) Hormone replacement therapy

3) Bisphosphonate medications such as Fosamax & Actonel

Osmosis Osteoporosis

Osteomalacia & Rickets

Diseases of Vitamin D and/or phosphate deficiency.

Rickets - affects children.
Osteomalacia - affects adults.
Symptoms: similar but more pronounced in the growing bones of children.
Fatigue, progressive muscular stiffness, & pain.
Skeletal deformities, such as bowed legs & loss of height.
Treatment: Adequate intake of Vitamin D, hormone Calcitonin

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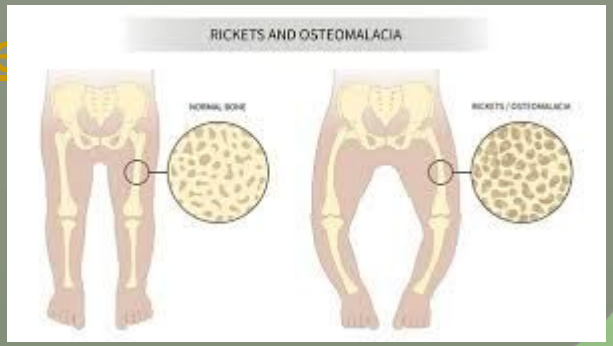


Pagets Disease

- Paget's disease (PD) (osteitis deformans)
- **2nd most common metabolic bone disease**
- Characterised by a disturbance in bone modelling and remodelling due to an increase in osteoblastic and osteoclastic activity.
- more common in people of Anglo Saxon origin
- The overall prevalence of PD is 3–3.7% and increases with age. By the age of 90 years, the prevalence increases to about 10%.
- Although it is classified as a metabolic bone disorder, it does not quite fit the category as some bone remains unaffected & physiologically normal. It;s etiology is somewhat unclear.

Signs and Symptoms

- Bone pain, deformity, fracture, and arthritis
  - pain of Paget's disease is located in the affected bone, most commonly; spine, femur, pelvis, skull, clavicle, humerus.
- The symptoms depend on the bones affected and the severity of the disease
  - Enlarged bones can pinch adjacent nerves, causing tingling and numbness
  - Bowing of the legs can occur
  - Hip or knee involvement can lead to arthritis, limping, as well as pain and stiffness of the hip or knee
  - Headache, loss of vision, and hearing loss can occur when the bones of the skull are affected.

Associated conditions

- Other Metabolic and endocrine diseases associated with joint/bone pathologies or symptoms are:
  - Gout syndrome
  - Osteogenesis imperfecta
  - Diabetes mellitus
  - Hyperparathyroidism
  - Thyroid disease
  - Abnormal Growth Conditions such as Hypopituitarism, Gigantism or Acromegaly

Growth Disorders

- Any type of problem in infants, children, or teenagers that prevents normal growth, which may be congenital (genetic) or acquired (hormonal irregularities, nutritional deficiencies)
- Hormones are necessary for normal growth and development, they regulate:
  - body's growth
  - Metabolism
  - sexual development and function.

Continued

- Hormonal causes of growth disorders include
  - thyroid hormone deficiency (hypothyroidism)
  - growth hormone deficiency
  - hypopituitarism
  - or other hormone disorders.
- some growth problems are not necessarily growth disorders; normal variants of growth patterns include genetic short height and slow growth/delayed puberty
- Growth hormone deficiency involves the pituitary gland.(produces GH and other hormones) GH is necessary for normal growth and development in children. In adults, growth hormone is needed to maintain the proper amounts of body fat, muscle, and bone.
- Growth hormone deficiency can occur at any age. In children growth will be slower than normal.In adults, low or absent growth hormone can also cause emotional symptoms, such as tiredness and lack of motivation.
- Hypopituitarism occurs if other pituitary hormones are absent or present in inadequate amounts

Cause

- low or absent secretion of GH from the pituitary gland
- May be present at birth (congenital/genetic)
- May develop after birth due to:
  - trauma, infections, radiation to the head, or tumors
- Adults with GH deficiency usually have a history of pituitary tumors treated with surgery or radiation
- In some cases, no cause can be identified. (idiopathic)

Acromegaly

- hormonal disorder that develops when the pituitary gland produces **too much growth hormone during adulthood** causing the bones to increase in size, including those of the hands, feet and face. Acromegaly usually affects middle-aged adults.
- **In children who are still growing, too much growth hormone can cause a condition called gigantism.** These children have exaggerated bone growth and an abnormal increase in height.
- If not treated promptly, acromegaly can lead to serious illness and even become life-threatening.

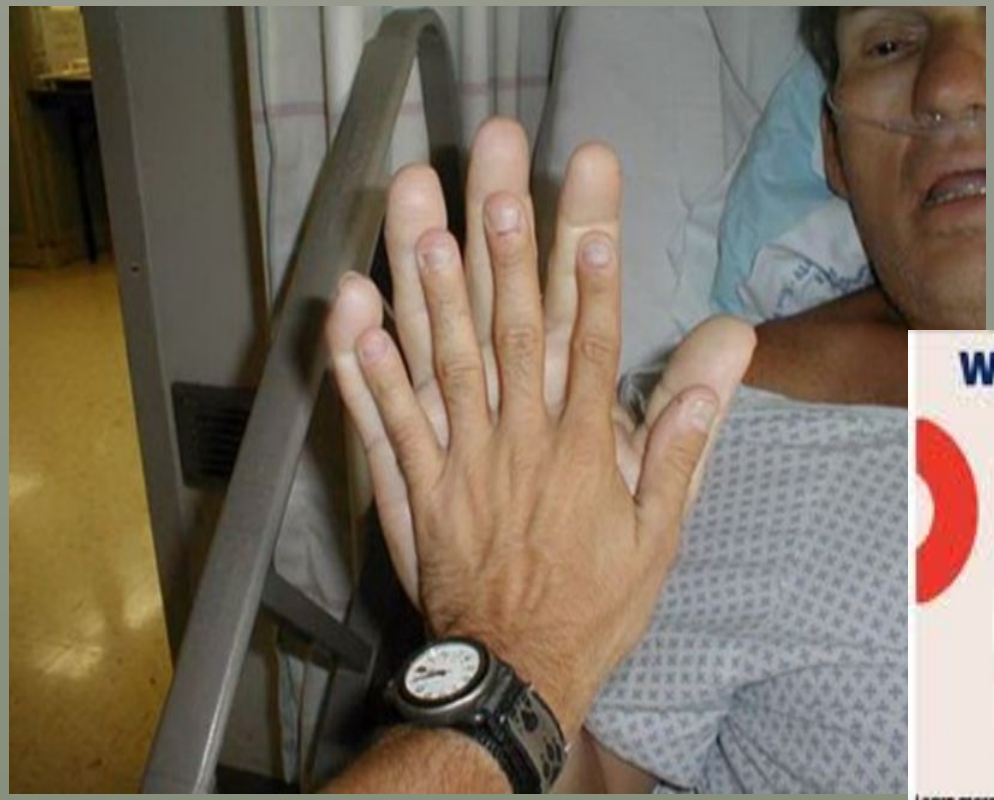
Acromegaly Signs and Symptoms

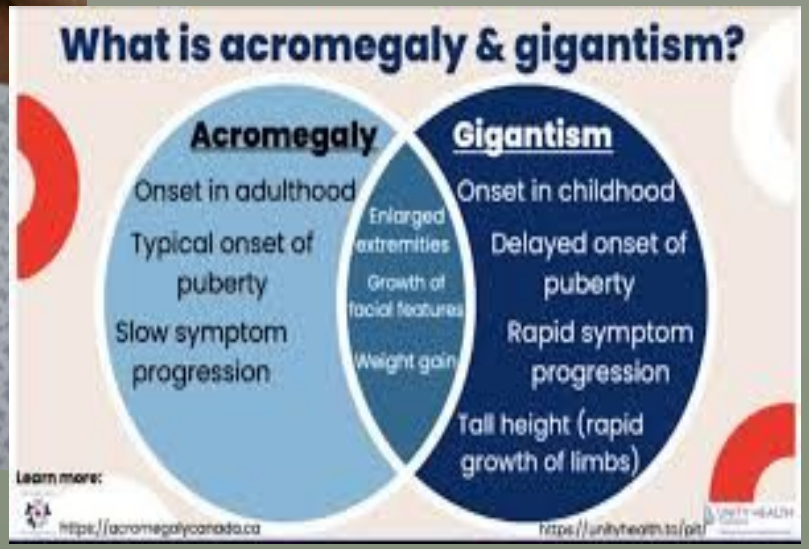
- One of the most common signs of acromegaly is enlarged hands and feet. People with this disorder often notice that they can no longer put on rings that used to fit and that their shoe size has progressively increased.
- Acromegaly may also cause gradual changes in the shape of the face, such as a protruding lower jaw and brow, an enlarged nose, thickened lips. Acromegaly may produce the following signs and symptoms, which can vary from one person to another:
  - Enlarged hands and feet
  - Coarsened, enlarged facial features
  - Coarse, oily, thickened skin
  - Excessive sweating and body odor
  - Small outgrowths of skin tissue (skin tags)
  - Fatigue and muscle weakness
  - A deepened, husky voice due to enlarged vocal cords and sinuses
  - Severe snoring due to obstruction of the upper airway
  - Impaired vision
  - Headaches
  - Enlarged tongue
  - Pain and limited joint mobility
  - Menstrual cycle irregularities in women
  - Erectile dysfunction
  - Enlarged liver, heart, kidneys, spleen and other organs
  - Increased chest size (barrel chest)

Complications

- Hypertension
- Cardiovascular disease, particularly enlargement of the heart (cardiomyopathy)
- Osteoarthritis
- Diabetes mellitus
- Polyps on the lining of the colon
- Sleep apnea
- Carpal tunnel syndrome
- hypopituitarism
- Uterine fibroids, benign tumors in the uterus
- Spinal cord compression
- Vision loss

Acromegaly




What is acromegaly & gigantism?
Acromegaly
Onset in adulthood
Typical onset of puberty
Slow symptom progression
Enlarged extremities
Growth of facial features
Weight gain
Gigantism
Onset in childhood
Delayed onset of puberty
Rapid symptom progression
Tall height (rapid growth of limbs)
Learn more:
https://acromegalycanada.ca
https://unityhealth.to/pit/

Connective Tissue and Joint pathology

- Arthritis
  - Types of Arthritis:
    - **Osteoarthritis** - Degenerative, affects weight-bearing joints
    - **Rheumatoid arthritis** - Autoimmune, affects small joints symmetrically
    - **Juvenile arthritis**
    - **Psoriatic arthritis**
    - **Gout**
    - **Septic Arthritis**

Osteoarthritis (OA)

- AKA degenerative joint disease (DJD) or degenerative arthritis
- most common chronic condition of the joints
- affecting approximately 27 million Americans
- Occurs in people of all ages, OA is most common in people older than 65.
- Breakdown in the cartilage covering the ends of bones where they meet to form a joint. Bones become exposed and rub against each other
- Deterioration of cartilage also affects the shape and makeup of the joint so that it no longer functions smoothly.
- Other problems can occur inside the joint as cartilage breakdown affects the joint components. Fragments of bone or cartilage may float in joint fluid, causing irritation and pain. Spurs, or osteophytes, can develop on the ends of the bones, damaging surrounding tissues and causing pain. Fluid inside the joint may not have enough of a substance called hyaluronan, which may affect the joint's ability to absorb shock. And although inflammation is not a main symptom of osteoarthritis, it can occur in the joint lining in response to the cartilage breakdown

Symptoms & Causes

- vary, depending on which joints and how severely they are affected
- most common symptoms are:
  - Stiffness, particularly first thing in the morning or after resting, and pain.
- most commonly affected joints are
  - lower back,
  - hips
  - knees
  - Feet
- Other commonly affected joints are:
  - neck and fingers, including the thumb base (making it difficult to grasp and hold objects, or to do delicate tasks
- Has no single, specific cause.
- Progressive and when it becomes severe it can severely affect an individuals day-to-day activities.

Risk Factors

- Age: OA is a degenerative condition
- Family History/Genetics
- Childhood hip diseases including:
  - DDH, Legg-Calve-Perthe's Disease and slipped capital femoral epiphysis
- Acquired conditions; ex: avascular necrosis can result in hip arthritis if left untreated or if treatment fails.
- Severe trauma. Fractures or traumatic dislocations
- Obesity

OA & the SI Joint

- OA is the most common cause of SI joint dysfunction.
- Pregnancy is a common cause of SI joint dysfunction. During pregnancy, hormones are released in the woman's body that allows ligaments to relax in order to prepare the body for childbirth. Relaxation of the ligaments holding the SI joints together allows for increased motion in the joints and can lead to increased stress and abnormal wear. The additional weight and altered gait associated with pregnancy also places additional stress on the SI joints.
- Any condition that alters the normal walking pattern places increased stress on the SI joint, including:
  - leg length discrepancy
  - pain in the hip, knee, ankle, or foot
- Patients with severe pain in the lower extremity often develop problems with either the lumbar spine or SI joints. In most cases, if the underlying problem is treated the associated lumbar spine or SI joint dysfunction will also improve.

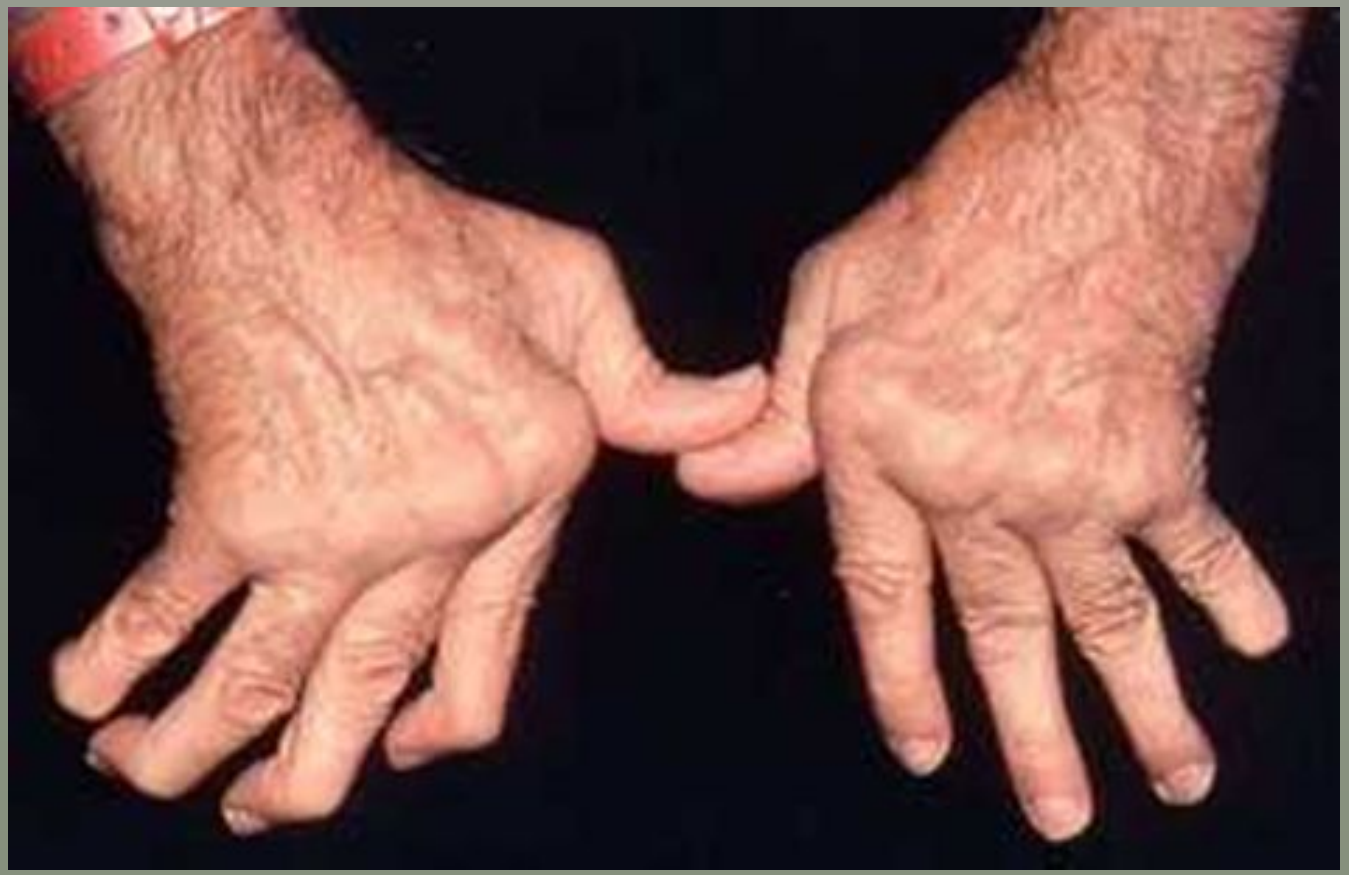
Rheumatoid Arthritis (RA)

- Though RA affects the joints; it's a disease of the immune system. RA causes the immune system to go awry and mistakenly attack healthy cells such as the synovium. As a result, fluid builds up in the joints, causing pain and inflammation.
- Over time this can wear away the cartilage and erode bone, causing a lack of function and mobility. In most people, the inflammation usually becomes systemic, affecting organs such as the skin, heart and lungs.
- RA most commonly affects the hands, feet, wrists, elbows, knees and ankles.
- Joint involvement is usually symmetrical
- no cure - its symptoms often come and go
- Affects an estimated 1.5 million Americans
  - 3 x more women than men
  - Usual age for adult onset is between 40 and 60 years, can begin at any age

Diagnoses & Signs and Symptoms

- Medical history
- Physical examination:
  - look for swelling, warmth, tenderness and limited motion in joints, or nodules under the skin.
- Signs and symptoms:
  - Regular morning joint stiffness
  - Persistent joint pain that does not improve
  - Joint pain that is getting worse over time
  - Joints that are swollen, red, hot or tender to the touch
  - Joint pain accompanied by fever
  - Several affected joints
  - Joint problems that affect ability to move or function
  - The "rheumatoid factor" is an antibody that can be found in the blood of 80% of people with rheumatoid arthritis.

RA in the hands



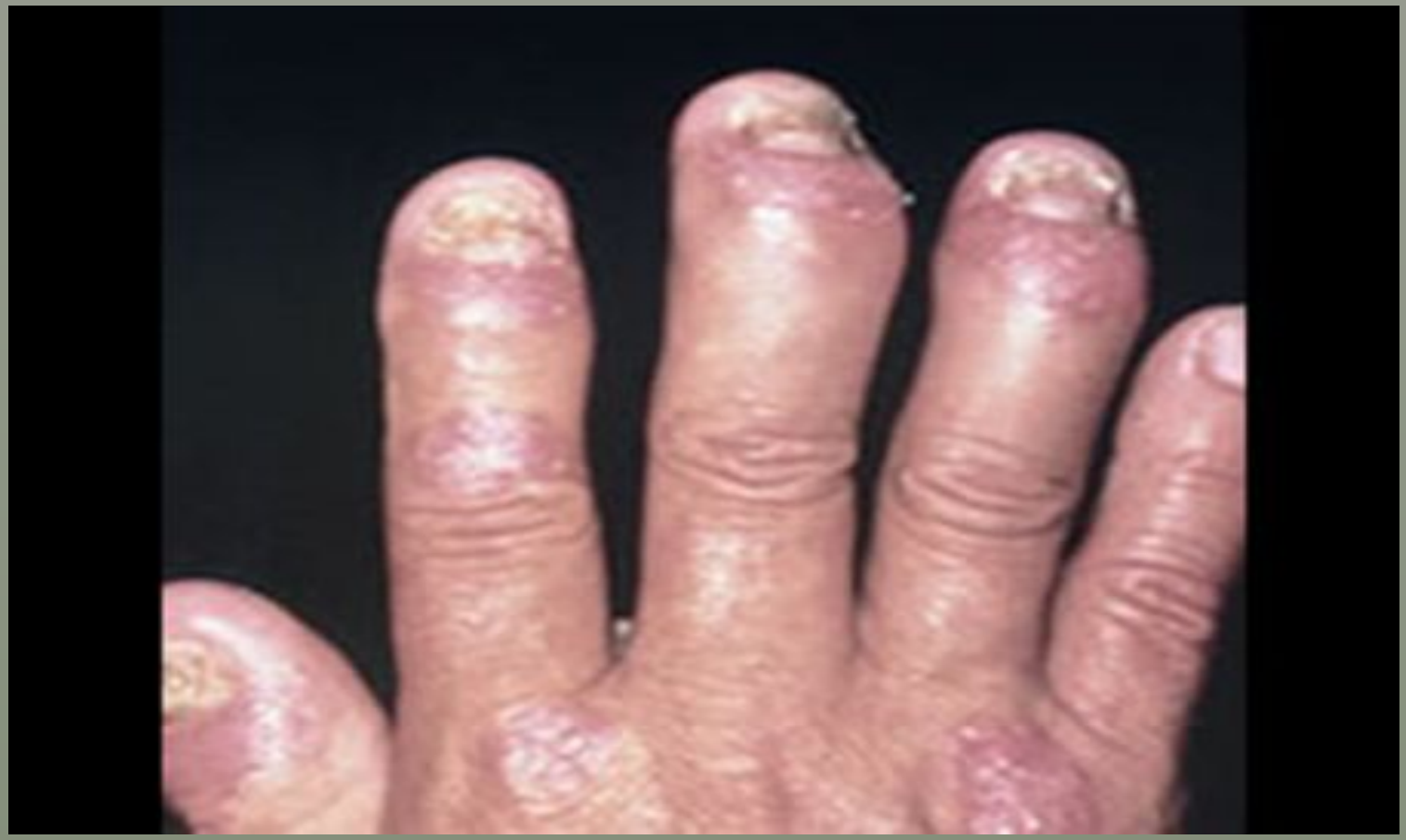
Juvenile Idiopathic Arthritis (JIA)

- Begins before age 16 and involves swelling in one or more joints lasting at least six weeks.
- Symptoms include:
  - muscle and soft tissue tightening
  - bone erosion
  - joint misalignment
  - changes in growth patterns
- Not all symptoms are shared by all children with the disease
- Symptoms can change from day to day
- Diagnosis of JIA is based on physical exam, lab tests and medical history. In addition to watching for symptoms for at least six weeks

Psoriatic Arthritis (PsA)

- autoimmune disease
- a form of inflammatory arthritis that can cause pain, swelling and sometimes damage to any joint in the body. It typically appears in people who have psoriasis, a chronic disease characterized by a scaly, reddish skin rash that usually appears on the elbows, knees and scalp
- most commonly associated with joints, it is a systemic condition,over time it can affect multiple joints and even organs.

Psoriatic Arthritis



Gout

- characterized by sudden, severe attacks of pain, redness and tenderness in joints, often the joint at the base of the big toe.
- can affect anyone. Men are more likely but women become increasingly susceptible to gout after menopause
- An acute attack of gout can wake the patient up in the middle of the night due to pain. The affected joint is hot, swollen and so tender that even the weight of the sheet on it may seem intolerable.
- Treatable
- Signs and symptoms include:
  - intense joint pain most severe within first 12 -24 hours
  - usually affects the joint of big toe, but it can occur in the feet, ankles, knees, hands and wrists
  - Lingering discomfort. After the most severe pain subsides, some joint discomfort may last from a few days to a few weeks. Later attacks are likely to last longer and affect more joint
  - Inflammation and redness

Causes Complications & Risk Factors

- urate crystals accumulate in the joint, creating sharp, needle-like urate crystals in a joint or surrounding tissue that cause intense pain, inflammation and swelling. Urate crystals can form when there is high levels of uric acid in the blood. The body produces uric acid when it breaks down purines as well as in certain foods, such as organ meats, anchovies, herring, asparagus and mushrooms.
- Urate crystals may collect causing kidney stones,
- Factors that increase the uric acid level in the body include:
  - Lifestyle factors. Excessive alcohol use — generally more than two drinks a day for men and more than one for women — increases the risk of gout.
  - High Fructose Corn Syrup consumption
  - Medical conditions. Certain diseases and conditions. These include untreated hypertension, chronic conditions (diabetes), hyperlipidemia, arteriosclerosis
  - Certain medications. The use of thiazide diuretics (treat hypertension , low-dose aspirin, anti-rejection drugs can increase uric acid
  - Family history of gout.
  - Age and sex

Osmosis Gout

Autoimmune Diseases affecting Connective tissues & Joints

Rheumatoid Arthritis
Psoriatic Arthritis
Juvenile Arthritis
SLE
Polymyositis
Dermatomyositis

Systemic Lupus Erythematosus

See Immunologic Dysfunctions' for description

- No two cases of lupus are exactly alike
- Signs and symptoms may come on suddenly or develop slowly, may be mild or severe, and may be temporary or permanent. Most people with lupus have mild disease characterized by flares.
- The most common signs and symptoms include:
  - Fatigue and fever
  - Joint pain, stiffness and swelling
  - Butterfly-shaped rash on the face that covers the cheeks and bridge of the nose
  - Skin lesions that appear or worsen with sun exposure
  - Raynaud's phenomenon
  - Shortness of breath
  - Chest pain
  - Dry eyes
  - Headaches, confusion, memory loss

Risk Factors

- Sex - more common in women.
- Age - most often diagnosed between the ages of 15 and 40
- Race - more common in African Americans, Hispanics and Asians.

Complications

- Kidneys -serious kidney damage, and kidney failure is one of the leading causes of death among people with lupus. S & S may include generalized itching, chest pain, nausea, vomiting and edema
- Brain - causes headaches, dizziness, behavior changes, hallucinations, strokes or seizures, memory problems and may have difficulty expressing their thoughts.
- Blood and blood vessels - blood problems, including anemia and increased risk of bleeding or blood clotting, vasculitis
- Lungs - pleurisy
- Heart.- inflammation of the heart muscle, arteries or pericarditis, cardiovascular disease and heart attacks increases
- Infection - more vulnerable to infection because both the disease and its treatments weaken the immune system. Infections include urinary tract infections, respiratory infections, yeast infections, salmonella, herpes and shingles.
- Bone tissue death - avascular necrosis, The hip joint is most commonly affected.
- Pregnancy complications - increased risk of miscarriage, preeclampsia and preterm birth.

Polymyositis

- uncommon inflammatory disease that causes muscle weakness affecting both sides of the body.
- can make it difficult to climb stairs, rise from a seated position, lift objects or reach overhead
- most commonly affects adults in their 30s - 50s
- more common in African Americans than in Caucasian's
- women are affected more often than men
- Develops gradually, over weeks or months
- The muscle weakness involves the muscles closest to the trunk, such as those in hips, thighs, shoulders, upper arms and neck. The weakness affects both the left and right sides of the body
- The exact cause is unknown, but the disease shares many characteristics with autoimmune disorders

Complications

- Dysphagia - If the esophageal muscles are affected, which in turn may cause weight loss and malnutrition.
- Aspiration pneumonia - Difficulty swallowing may also cause aspiration of food or liquids, including saliva, into the lungs, which can lead to pneumonia.
- Breathing problems - If the chest muscles are affected by the disease, the patient may experience breathing problems, such as shortness of breath or, in severe cases, respiratory failure.
- Calcium deposits - Late in the disease, deposits of calcium can occur in the muscles, skin and connective tissues (calcinosis).

Associated Conditions

- Raynaud's phenomenon
- Other connective tissue diseases such as lupus, rheumatoid arthritis, scleroderma and Sjogren's syndrome, can occur in combination with polymyositis.
- Cardiovascular disease muscular walls of the heart to become inflamed (myocarditis). In a small number of people who have polymyositis, congestive heart failure and heart arrhythmias may develop.
- Lung disease - A condition called interstitial lung disease may occur with polymyositis. Interstitial lung disease refers to a group of disorders that cause scarring (fibrosis) of lung tissue, making lungs stiff and inelastic. Signs and symptoms include a dry cough and shortness of breath.

Other causes of Connective Tissue and Joint Disease

Scleroderma
Charcot's Joints (Neurogenic Arthropathy)
Hemarthrosis

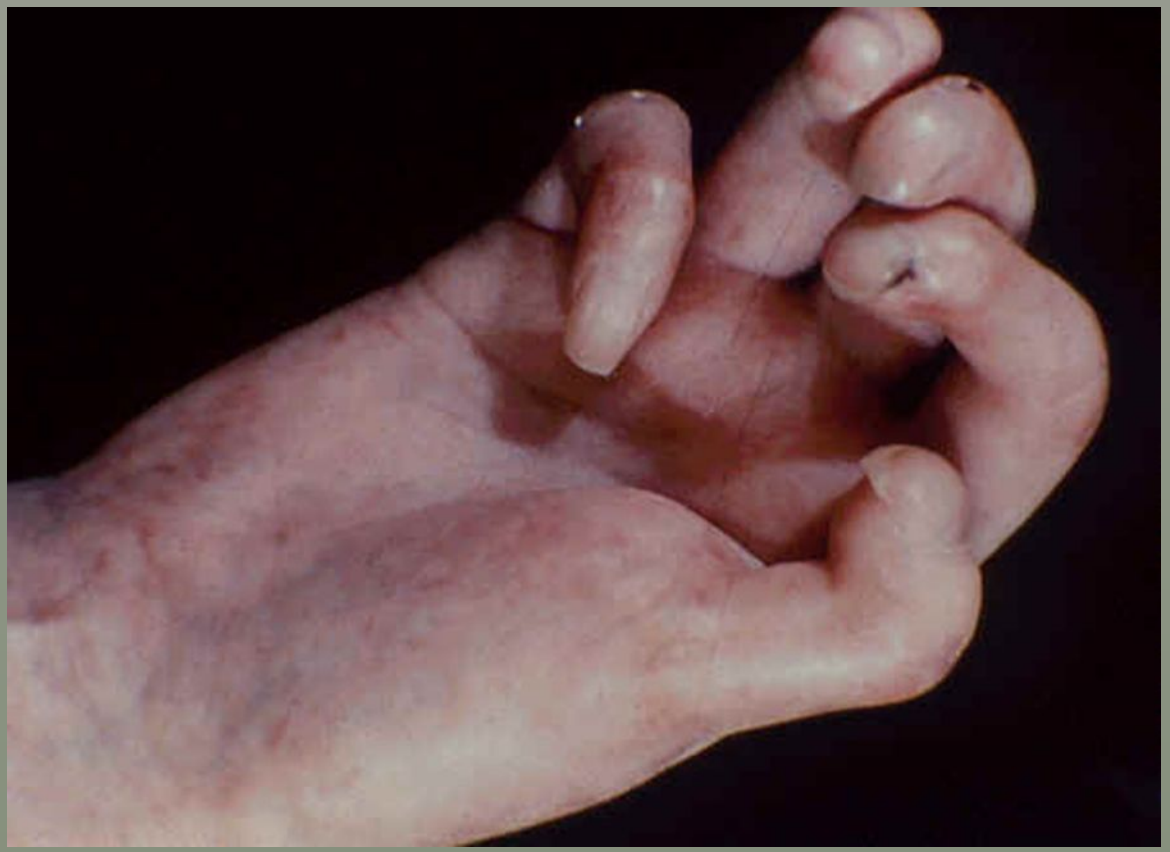
Scleroderma

- A group of rare diseases that involve the hardening and tightening of the skin and connective tissues.
- Caused by an overproduction and accumulation of collagen in body tissues.
- In some scleroderma affects only the skin. But can also harm blood vessels, internal organs and the digestive tract
- Affects women more often than men
- most commonly occurs between the ages of 30 and 50
- No cure for scleroderma, but treatments can ease symptoms and improve quality of life.

Signs & Symptoms

- Skin -hardening and tightening of patches of skin, which may be shaped like ovals or straight lines. number, location and size of the patches vary by type. Skin can appear shiny and movement of the affected area may be restricted.
- Fingers or toes - exaggerated response to cold temperatures or emotional distress, which can cause numbness, pain or color changes in the fingers or toes. Called Raynaud's phenomenon.
- Digestive system - acid reflux, which can damage the section of esophagus nearest the stomach, some people with scleroderma may also have problems absorbing nutrients if their intestinal muscles aren't moving food properly through the intestines.
- Heart, lungs or kidneys. Rarely, scleroderma can affect the function of the heart, lungs or kidneys. These problems can become life-threatening.

Scleroderma Hand



Neurogenic Arthropathy (Charcot Joints)

- **A rapidly destructive arthropathy due to nerve damage resulting in impaired pain perception and position sense. The nerve damage can occur from many causes, most commonly diabetes and stroke.**
- Symptoms include joint swelling, effusion, deformity, and instability. Pain may be disproportionately mild due to the underlying neuropathy.
- Diagnosis requires x-ray
- Impaired deep pain sensation or proprioception affects the joint's normal protective reflexes, often allowing trauma (especially repeated minor episodes) and small periarticular fractures to go unrecognized. Increased blood flow to bone from reflex vasodilation, resulting in active bone resorption, contributes to bone and joint damage. Each new injury sustained by the joint causes more distortion as it heals. Hemorrhagic joint effusions and multiple small fractures can occur, accelerating disease progression. Ligamentous laxity, muscular hypotonia, and rapid destruction of joint cartilage are common, predisposing to joint dislocations, which also accelerate disease progression. Advanced neurogenic arthropathy can cause hypertrophic changes, destructive changes, or both.

Continued

- **Arthropathy does not usually develop until years after onset of the neurologic condition but can then progress rapidly and lead to complete joint disorganization in a few months**. Pain is a common early symptom. However, because the ability to sense pain is commonly impaired, the degree of pain is often unexpectedly mild for the degree of joint damage. A prominent, often hemorrhagic, effusion and subluxation and instability of the joint are usually present during early stages. Acute joint dislocation sometimes occurs also.
- During later stages, pain may be more severe if the disease has caused rapid joint destruction (eg, periarticular fractures or tense hematomas)
- During advanced stages, the joint is swollen from bony overgrowth and massive synovial effusion. Deformity results from dislocations and displaced fractures. Fractures and bony healing may produce many loose pieces of cartilage or bone that can slough into the joint, causing a coarse, grating, often audible crepitus usually more unpleasant for the observer than for the patient. The joint may feel like a "bag of bones."

Continued

- knee and the ankle are most often affected
- Distribution depends largely on the underlying disease.
  - ex - diabetes mellitus affects the foot and ankle, Syringomyelia commonly affects the spine and upper limb joints, especially the elbow and shoulder
- Frequently, only one joint is affected and usually no more than two or three (except for the small joints of the feet), in an asymmetric distribution.
- Infectious arthritis may develop with or without systemic symptoms particularly with diabetes. Structures such as blood vessels, nerves, and the spinal cord can become compressed due to the tissue overgrowth.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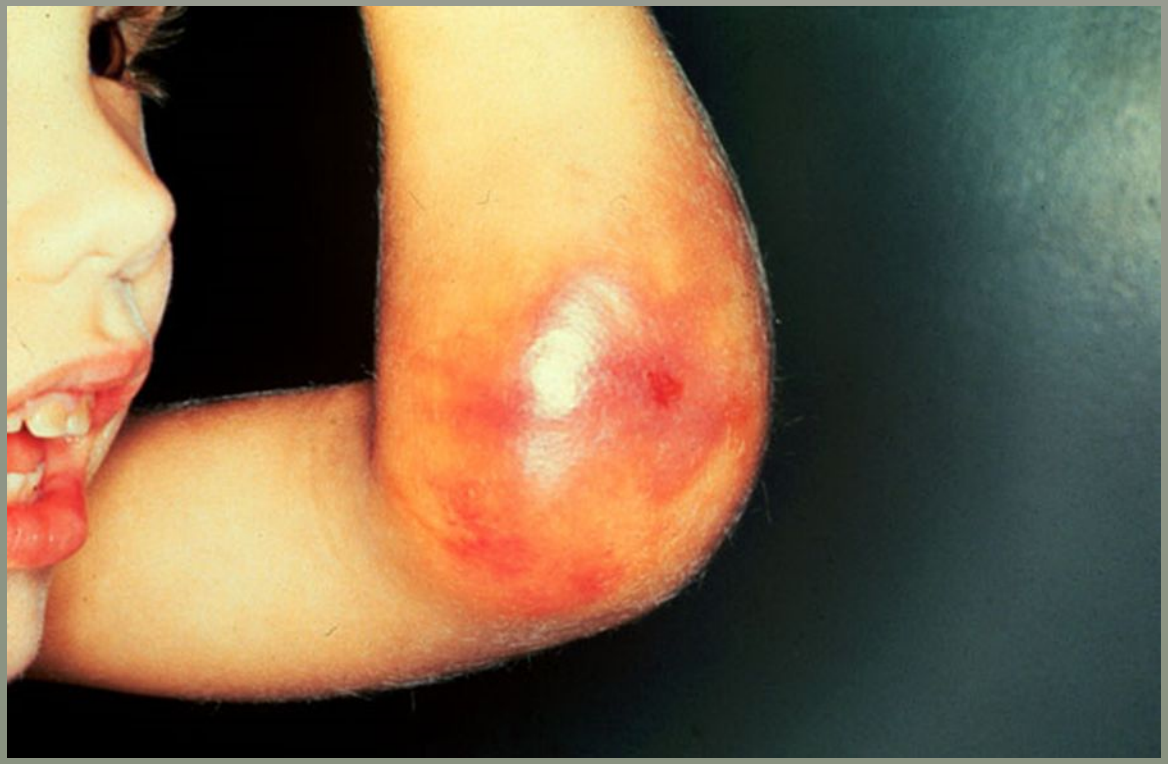
Hemarthrosis

- **Bleeding into a joint. May be caused by: Coagulation disorders such as Hemophilia, Use of Anti-coagulation medications such as Warfarin, or Trauma to asn otherwise healthy joint.**
- In hemophilia, joint bleeding usually begins before the age of five and tends to recur repeatedly during childhood in response to even minor injury. The most common sites are the knees, elbows, and ankles, but any joint can be involved.
- Acute hemarthrosis usually results in marked local inflammation and joint symptoms that can last for days to weeks.
- Approximately one half of patients with hemophilia develop chronic deformities in one or more joints. Some of them develop a chronic progressive synovitis, restricted to one or a few joints, which clinically resembles rheumatoid arthritis.
- In chronic cases there is marked synovial membrane hyperplasia, destruction of articular cartilage, and erosions of subchondral bone. This chronic progressive pattern probably results from a low level of continuous or intermittent bleeding into involved joints. joint fluid, in chronic cases, usually contains blood and very high levels of leukocyte-derived proteases. Other musculoskeletal manifestations of hemophilia include bleeding into muscle and bone.

Continued

- Symptoms of hemarthrosis are usually first noticed during infancy or childhood. milder forms of hemophilia may not have symptoms until later in life. Symptoms of bleeding into a joint include:
  - Warmth and/or tingling in the joint during the early stages of hemarthrosis. This is called an aura. If bleeding is not treated, it can progress to severe pain.
  - Swelling and inflammation in the joint, caused by repeated episodes of bleeding. If episodes continue, it may lead to chronic pain and destruction of the joint.
  - An infant's or child's reluctance to move an arm or leg because of bleeding into an affected joint, often first noticed when a child begins to walk.
- It usually follows injury but occurs mainly in patients with a predisposition to hemorrhage such as those being treated with warfarin and patients suffering from hemophilia. It can be associated with knee joint arthroplasty.



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