Musculoskeletal

(Med I, Block 5, MS)
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Objectives:

Following the 50 minute lecture, the student will be able to:

- Explain in course details the process of intramembranous and endochondral bone formation and muscle formation
- Define interzonal mesenchyme
- Explain the role of the Apical Ectodermal Ridge, the Progress Zone, and the Zone Polarizing Activity in upper limb formation
- Explain the role of Shh and Hox genes in the formation of the upper limb
- Explain the embryological basis of the dermatomes
- Describe the embryological basis of achondroplasia, syndactyly, talipes equinovarus, Erb’s Palsy, and congenital hip dysplasia

Induction:
- Condensation – somites from mesoderm

Formation of Bone:
- Intramembranous = e.g. ilium, parietal, scapula – direct from mesodermal derivatives
- Endochondral = bones of the limbs – cartilage model – \textbf{WHAT TYPE OF CARTILAGE?}

Formation of Muscle:
- Differentiation of myoblasts from mesoderm - hyperplasia, fusion
- \textless{} 26 weeks \textit{in utero} – Type I (slow twitch) - mostly hyperplasia
- \textgreater{} 26 weeks \textit{in utero} – Type II (fast twitch) - increasing hypertrophy
- Full term
  - \textbf{WHAT PERCENT OF TOTAL NUMBER OF ADULT FIBERS?}
  - 25\% total body weight
- Postnatal – 1-5y achieve adult number

Formation of Joints:
- Interzonal mesenchyme (mesoderm cells between developing bones)
- Differentiation and condensation of cells
  - Fibrocytes – fibrous joint – \textbf{EXAMPLE:}_______________________________
  - Cartilage +/- fibrocytes – fibrous joint – \textbf{EXAMPLE:}________________________
  - Synovial Joint
    - Trilaminar (three layer structure) –
    - Adjacent to bone ends – articular cartilage
    - \textbf{WHAT DOES OUTER LAYER FORM?}
    - Inner layer – Cavity (apoptosis), ligaments, meniscus
Formation of Limb/Digits:
- Induction
- Control Zones
  - Apical Ectodermal Ridge
    - stimulates lengthening (proximodistal) of limb
    - maintains the signals for patterning
  - Progress Zone
    - pool of undifferentiated mesenchyme cells
    - *WHAT KEEPS THIS ZONE UNDIFFERENTIATED?*
  - Zone Polarizing Activity
    - “compass”
    - controls anterior-posterior axis
- Genes – control zone
  - Sonic Hedgehog (Shh) gene - ZPA
    - *TO WHAT ORIENTATION DOES SHH ESPECIALLY CONTRIBUTE?*
  - HOX genes
    - Limb gradient (proximodistal development)
- Digit Formation
  - Apoptosis
- Innervation
  - Myotome (somites) cells “pull” nerves from adjacent developing spinal cord
    - Dermatome – distribution related to limb origin, lengthening, rotation

Congenital Defects:
- Achondroplasia
  - Fibroblast growth factor (FGF), increased age of father
  - *WHAT ARE THE SKELETAL CHANGES?*
- Syndactyly
  - *WHICH GENE CAN BE INVOLVED IN SOME SPECIFIC SYNDACTYLY CASES?*
- Talipes
  - Deformation (versus a defect)
  - *WHAT IS TYPICAL TREATMENT?*
- Erb’s Palsy
  - Shoulder dystocia
  - *WHAT IS INJURED?*
- Developmental Hip dysplasia
  - Joint laxity relating to femoral head/acetabulum/joint structure
  - *WHAT IS A BARLOW TEST?*
SAMPLE EXAM QUESTIONS:

Which of the following contributes most to the normal pinkie-to-thumb orientation of the hand:

a. Lack of HOX genes in the developing limb  
b. Gradient of Shh gene products  
c. Fibroblast growth factor (FGF)  
d. Orientation of the nerves in the brachial plexus  
e. Formation of the interzonal mesenchyme

The Apical Ectodermal Ridge is responsible for which one of these limb developmental events:

a. Proximodistal lengthening  
b. Pinkie-to-thumb orientation  
c. Formation of the joint meniscus  
d. Formation of the joint capsule  
e. Primary centre for endochondral bone formation and ossification
OBJECTIVES:
1. To be able to define and classify scoliosis.
2. To be able to appreciate the importance of a good history and the physical signs of scoliosis.
3. To be able to order investigations appropriate to scoliosis and spondylolysis.
4. To understand the principles of conservative and surgical management.

REFERENCES: Essentials of Surgical Specialties, Ch 7, Pg 264-65.
Textbooks of Disorders and Injuries of the Musculoskeletal System - Salter

Scoliosis:
Definition – 3D deformity of the spine with frontal plane deformity of the spine > 10 degrees.
Most common back deformity.

⇒ Risks of progression: cosmetic deformity, pain, cardiorespiratory compromise from rib cage restrictive dysfunction.

Classifications:
a. Acquired:
   a. Trauma: including infections
   b. Iatrogenic: post-radiation, thoracoplasty ⇒ growth arrest
   c. Neoplastic
b. Neuromuscular:
   a. UMN: cerebral palsy, spinal cord injury, paralysis
   b. LMN: spina bifida, spinal muscular atrophy
   c. Myogenic: muscular dystrophies, such as Duchenne’s MD
-- All three types are very likely to progress; almost all will need surgery to prevent cardiorespiratory compromise.

c. Secondary:
   a. Leg length discordance, e.g. femoral shortening
   b. Muscle Spasm

d. Idiopathic:
   a. Infantile:
   b. Juvenile: True scoliosis in 2-3% of children
   c. Adolescent: *** most common

Idiopathic Adolescent Scoliosis:
- Most common but < 10% will need any treatment.
- Etiology – research suggests neurological basis.
- F > M – 8:1, progressive requiring surgery.
- Right thoracic curve most apt to progress.
- Risk factors for progression are gender, curve location, magnitude, age/ maturity at presentation.
- Low risk factors: – family history, amount of lordosis, kyphosis.

Investigations: A detailed history and physical exam (i.e. curve, rib hump) is most important.

Concerns on History:
- Idiopathic or not.
- Age at onset, including menarchal or not.
- Rate of progression.
- Family History of scoliosis and need for treatment.
- Pain.

Imaging Techniques
- 3 foot standing PA spine,
- wrist x-ray for bone age
- Risser sign: degree of ossification of iliac wing.

Treatment:
Rule of thumb – If > 6 months’ growth remaining:
- 10 – 20 degrees – observe
- 20 – 40 degrees – brace (TLSO, Milwaukee)
- > 50 degrees – think surgery (multilevel fusion)
Congenital Scoliosis:

i. **Classification:**
   1. Failures of segmentation:
      - Wedge vertebrae
      - Hemivertebrae
   2. Failures of formation:
      - Congenital bars
      - Block vertebrae
   3. Both + ribs

Associated organ system involvement
   - Renal: 20%
   - CVS: 10%

Progression
   - 25% static
   - 25% mild progression
   - 50% progress significantly

Treatment – Brace vs. surgery (role of MRI)

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**Spondylolysis & Spondylolisthesis:**

**Definition:**

- **Lysis** – a defect (congenital or acquired) of the pars interarticularis
  - usually L5 on S1
- **Listhesis** – a slippage of one vertebra on another (forward or backward)
  - usually L5 on S1

High Risk:  Gymnastics/Football – role of extension.

Physical Exam:  Flat back, palpable defect, tight hamstrings, decreased SLR

Treatment:  Lysis – no pain – observe

  Lysis – pain – brace or surgery

  Listhesis:  
    - Minor with no pain  →  observe
    - Minor with pain  →  brace, spine stabilization exercises
    - Major +/- pain  →  surgery – fusion.
      (= >50%+ slippage)

Bracing options:
TLSO (thoracolumbosacral orthosis – custom plastic clamshell)
CTLSO (cervical TLSO) – if curve incorporates high thoracic or cervical level

Radiography Review:

Oblique x-rays: “Scottie dog with collar” -- lysis
“decapitated Scottie dog” -- listhesis

Oblique: Lateral:

Case:
Progressive Right thoracic scoliosis in 16 year old male.
Ominous?

All images from WebMD.

Yes – MRI – holocord astrocytoma
Objectives: Through an interactive discussion of the following case scenarios, the participant will gain further knowledge and expertise in the presentation, diagnosis and management of the following upper extremity issues: a) anterior and posterior glenohumeral dislocations; b) compartment syndromes in the arm; c) FOOSH injuries, including scaphoid and distal radial-ulnar fractures.


SHOULDER DISLOCATION

Mom? Is that you? Thanks for accepting the call.

Yeah, hi to you too. I’ve got some good news. This afternoon I thought I was going to have to drop out of med school but the doctor says I am going to be OK. Mom? I can’t hear you.

Hi, Dad. Is Mom OK?

“Yes, I’m at the ski slope. Remember that I told you I was going skiing for several days at the beginning of Christmas vacation...

What happened? That’s what I was trying to tell Mom. I dislocated my shoulder, but I’m going to be OK.

No, it’s my left shoulder, which is great because I can still wrist, but I can’t drive my stickshift. Can you come get me?

OK, I’ll slow down. I was coming in for lunch, and you know how Bonzai levels out when you get close to the lift. I caught an edge right there and just flipped. It was all over before I knew it, and I was lying there in a crumpled heap. Every time I tried moving my left shoulder, wow! I’ve never had pain like that before.

No, I picked up my skis and poles and walked to the first aid station, holding my left arm still. They took me to the hospital.

Hi, Mom, you’re on the line too? Didn’t mean to scare you. More good news, Mom. They didn’t have to cut off the sweater you knitted for me. Dr. Rogers was great. Very gentle and thorough. I told him I was a med student interested in orthopedics, and he explained everything to me. He says that as I fell, I probably reached out instinctively and forced my shoulder into abduction and external rotation. He checked me over and compared the shape of one shoulder with the other. He knew right then what was wrong. It’s a common injury for people my age, especially during ski season. He checked my pulse as well as the sensation and strength in my hand. And he also checked the sensation over the outside of my shoulder. Sometimes a nerve can get stretched from the dislocation, and the deltoid or other muscles can be paralyzed.

No, Mother. I am not paralyzed. I’m standing at a phone booth in the hospital lobby.
The x-rays were fine, Dad. Just a garden-variety anteroinferior dislocation. No greater tuberosity or humeral neck fracture. Once he saw the x-rays he gave me some Demerol and reduced the dislocation by having me lie prone on the stretcher with my left arm dangling down over the side. Dr. Rogers said that was the safest way. In a few minutes, one of his associates pulled down the arm while he put his thumbs in my armpit and pushed the head back in place. Clunk, I felt better right away.

Uh huh. I am in a sling. He told me not to abduct or externally rotate my shoulder for at least four weeks. He said recurrent dislocation is the most common complication in my age group. I only need to let the tissues heal a bit. In people over 30, stiffness is the biggest problem, so he gets them moving right away. Apparently in elderly people this same injury mechanism fractures the humeral neck rather than dislocating the joint because the bone becomes weaker than the ligaments. Since I’m interested in Ortho, he drew me a sketch showing how sometimes the thickened capsular attachment to the glenoid rim, the labrum, becomes detached.

Those types don’t heal so well and need to be reattached to prevent recurrences. Also, the lip of the glenoid sometimes punches a crater in the humeral head during the dislocation. This makes recurrences more likely too, but my x-ray looked OK to him. “Are you still there? Good. Maybe just to scare me into always using my sling, he told me about the various surgical treatments for recurrent dislocation. If the labrum is torn off, they can reattach it. Otherwise, there are various ways to tighten up the joint capsule and subscapularis tendon to reduce the laxity. Unfortunately, this may work a little too well and affect an athlete’s ability to throw. Good thing I’m right handed.

What? Yeah, you’re right. The Demerol’s probably making me a little high. If you’ll come get me, I can tell you the rest on the way home. I’m going to get something to eat, and then I’ll wait here in the lobby. Thanks.

Questions:
1. Describe an important principle outlined by this case. (Several possible)

2. What nerve about the shoulder did the doctor test for?

3. Describe an alternate method of shoulder reduction (for anterior dislocation).
SUPRACONDYLAR FRACTURE OF THE HUMERUS COMPARTMENT SYNDROME

Ladies and gentleman of the jury, after two days of testimony from multiple witnesses and the defendant, Dr. Good, I will now summarize the sequence of events leading to the pain, misery, disfigurement, and severe functional loss that are a fact of life for this young man sitting before you.

Although Mackenzie Witherspoon had a rough start in life, at 8 years of age he seemed to be adjusting well to his new foster home, that is, until one morning last year when he fell from the neighbor’s tree while borrowing several apples. Typical for such a headfirst fall, the lad wisely extended his hands to protect himself. Unfortunately, because of this instinctive and highly judicious act, Mackenzie’s right elbow was forced into hyperextension, causing the humerus to break distally. This is an injury the experts have told us is common in children and adolescents and is best described as a supracondylar fracture. Two complications are well known: malunion with alteration of the carrying angle and compartment syndrome with devastation of the flexor muscles in the forearm. We have found no fault with his neighbor’s emergency aid; splinting the limb in an extended position with folded newspaper and masking tape; and once he explained to us Mackenzie’s unwillingness to come to the hospital, we understood the reason for transporting him in the trunk of his car.

The radiologist described to us the initial x-ray findings typical for this injury: a short oblique fracture of the distal humerus with proximal and posterior displacement of the distal fragment.

From the anatomic sketches introduced earlier, it is obvious to everyone here that the sharp edge of the proximal fragment pressing anteriorly against the nearby brachial artery could compress, if not actually tear or cut it. Furthermore, swelling from the injury could contribute to arterial occlusion.

Next, we come to Dr. Goode’s emergency room notes, sparse as they were. Let me re-read Dr. Good’s initial evaluation.

*Filthy urchin, screaming with elbow pain. Hyperextension deformity right elbow. No obvious breaks in skin. No other injuries apparent. X-rays show displaced supracondylar fracture. Will try for closed reduction and possible percutaneous pinning in OR under complete general anesthesia.*

Three experts reviewed this marginally legible note with us, and none could find any notation of this unfortunate lad’s initial distal neurovascular status. Each emphatically stated that such an evaluation is the standard of care in all cases and critical in an injury such as this in which vascular complications are well known.

Next, we have Dr. Good’s operative note. He was kind enough to read it to us in its entirety and to explain the medical terminology. Let me review several key sentences.
The little bastard bit the anesthesiologist during an otherwise routine crash induction and endotracheal intubation....Even with the elbow in complete flexion and hinging the distal fragment on the intact posterior periosteum and triceps, the fracture remained unstable and marginally reduced. Therefore, I inserted several crossed Kirschner wires from the lateral and medial epicondyles to stabilize the fracture. At this point the radial pulse was noted to be absent. X-rays now revealed anatomic reduction of the fracture with one K-wire protruding slightly anteriorly. When this K-wire was withdrawn, the radial pulse could be palpated....a long arm splint was applied and the patient left the OR in satisfactory condition. He will be admitted for overnight observation.

Now let me draw your attention to the meticulous notes Nurse Skylark made that night:

**11:00 pm** Eight-year-old boy admitted from the recovery room following percutaneous pinning of right elbow fracture. Patient says elbow hurting more. Vital signs stable, right arm in splint. Left hand cooler than right, radial pulses equal. Sensation to fingertips intact. Full passive extension of digits causes mild discomfort at the fracture site. Generally disheveled. Washed face. Told me he could curse in six languages.

**2:00 am** Patient complains of increasing pain. VS stable, radial pulse intact. Gave prn acetaminophen dose twice, spat first dose out.

**3:15 am** Patient screaming with pain. VS unobtainable due to lack of cooperation. Fingers held in flexed posture, efforts at passive digital extension cause agonizing pain. Fingers cool, capillary filling OK, diminished pinprick sensation on fingertips. Called Dr. Goode. He asked whether radial pulse was intact. I checked and it was. He prescribed codeine.

**3:55 am** Pain worse. Dr. Goode notified

**4:20 am** Dr. Goode arrives. Boy is swearing and writhing in pain. Dr. Goode says he can feel radial pulse and not to worry. He splits outer portion of the bandage. No change in pain. Orders Demerol. Medical student asks if she can do forearm compartment pressure study. Dr. Good says OK.

**4:30 am** Medical student wakes Dr. Goode at nurses’ station and reports forearm pressure of 85 mmHg.

**4:31 am** Dr. Goode transfers patient to gurney himself and runs to operating room

Gentlemen and ladies, the rest is sad, yet straight forward. The experts explained compartment syndrome, but let’s briefly review it. Bleeding into a limb adds volume, and in certain areas where the muscle fascia is unyielding, the added volume increases the pressure within the compartment. The muscles respond to the ensuing ischemia by releasing inflammatory mediators like histamine, and the problem is compounded because increased capillary permeability promotes edema. Also, capillary perfusion pressure may be reduced because of extra compartmental compression on the blood vessels. So, when the compartment pressure, which is rising, exceeds the capillary pressure, the enclosed tissues are no longer perfused.
The forearm and leg are particularly vulnerable to this devastating condition because of the tight fascial restraints surrounding the muscles in these locations. The physical findings are easy to remember – the five P’s – pain out of proportion, paresthesia, pallor and pulselessness. The first three are related to nerve ischemia and are early signs since the nerves are quite sensitive.

Any movement of the muscle heightens the pain; an observation Nurse Skylark noted when she tried to straighten the digits. The last two signs, pallor and loss of pulse, occur late and only after the compartment pressure exceeds systolic pressure. As you have learned, compartment pressure measurements are useful, particularly in comatose or otherwise uncooperative patients. As a temporizing measure, completely splitting all circumferential dressings to relieve any external compression may help, but emergency fasciotomy to allow for tissue swelling and restoration of capillary perfusion is critical. Split-thickness skin grafting several days later can then cover the swollen muscles. Unfortunately, when the fasciotomy is delayed, the prolonged ischemia and necrosis lead to permanent muscle and nerve damage, observations first noted Dr. Volkmann as long ago as 1881.

To Dr. Goode’s credit, his operative management of the supracondylar fracture resulted in anatomic alignment and this avoids the complication of a disfiguring malunion. The issue of a vascular complication, however, is a different matter. Dr. Goode admitted that his failure to perform and record an initial neurovascular examination while in the emergency room is defenseless. One can only surmise whether the subsequent vascular compromise was related to the fracture itself or to the manipulation and K-wire insertion in the operating room.

Dr. Goode’s adamant position has been that the radial pulse remained intact through the night, and therefore the boy’s pain was just surgical and not ischemic. Intuition and our expert witnesses, however, tell us that distal pulses remain intact until the compartment pressure approaches the systolic pressure exceeds capillary perfusion pressure. Dr. Goode, unfortunately, didn’t recognize that pain is the most sensitive of the five P’s; and if he had heeded Nurse Skylark’s initial warning about young Mackenzie’s spiraling pain, this young man could have led a productive and fruitful life. Now maimed, who knows what other tragedies await him in life. I rest my case.

Questions:
1. Describe an important principle outlined by the case (Several possible).

2. What is the most important symptom of an impending compartment syndrome?

3. Describe appropriate tests for median and ulnar nerve function below the elbow.
FALLS ON THE OUTSTRETCHED HAND (FOOSH):

After an unexpected ice storm, four patients in the emergency room, ages 6, 11, 17 and 70, give precisely the same history. Each slipped while walking across a parking lot and fell onto his or her outstretched hand – FOOSH! Each notes wrist pain and limited motion. The 17-year-old landed equally hard on both wrists and complains of pain bilaterally. Is it likely that they all have the same type of injury?

The distal neurovascular status of each is normal. You note tenderness in the wrist area of each patient. The following x-rays are available (to be distributed at the session)

Each patient sustained a different type of skeletal injury because the “weak link” in the skeleton varies with age.

In young children, the joint capsule is lax and supple, and the cartilaginous epiphyseal growth plate is stronger than the bone. Yet the bone too is somewhat supple, so an angularly directed force such as in a FOOSH injury merely buckles the bone, resulting in a torus fracture (known aliases – greenstick fracture, buckle fracture). In older children, the epiphyseal plate is weaker than the bone. FOOSH injuries now cause skeletal disruption through the growth plate. Five general patterns of epiphyseal injuries are recognized.

The epiphysis of the distal radius closes in girls at about age 16 and in boys several years later. The radius becomes stronger, and the adjacent scaphoid is now the weakest link, so a FOOSH injury can occur here in teenagers and young adults. Scaphoid fractures are not always easy to see on x-ray examination because the scaphoid lies obliquely to the planes of both posteroanterior and lateral wrist x-rays and because the fracture line through the scaphoid may lie in yet another plane.

Not every impact on the heel of the hand results in a fracture, of course. Ligamentous disruptions occur, especially at ages when the bone is strong. Major disruptions can result in lunate dislocation. Minor tearing of the supporting capsule is what is commonly known as sprain. Intervening degrees of disruption may be difficult to diagnose and may require special studies such as bone scan, arthrography, and arthroscopy.

With advancing age, osteoporosis gradually reduces the bone density of the distal radius, again rendering it the most vulnerable site in an FOOSH injury. This fracture of the one inch of the radius with apex palmar angulation of the distal fragment was first described by Abraham Colles in 1814 and bore his name for many years. (Irrelevant fact: It is the only article he ever wrote.) With recognition of other, slight different fracture patterns at the distal radius, the eponym war between hubristic surgeons became so furious that the trend now is just to call them all distal radius fractures and describe the exact pattern of the fracture lines.

Treatment of the greenstick fracture consists of casting to protect the limb from additional injury during the time required for healing. If there is no tenderness at the fracture site at three weeks, the cast may be left off.
Most epiphyseal fractures of the distal radius are type I or II. The desire for anatomic reduction must be balanced against the risk of further damage to the growth plate and possible growth arrest caused by multiple manipulations. With muscle relaxation and analgesia, longitudinal traction and wrist flexion can achieve anatomic or near anatomic reduction. A long arm cast (from axilla to mid palm) worn for four weeks usually suffices. Residual angular deformity adjacent to an open growth plate will correct spontaneously with further growth when the deformity is in the plane of major motion of the adjacent joint. Deformities in the two planes perpendicular to the motion will not correct. Slight under correction of the anterior apex angulation in this fracture will correct spontaneously with further growth, so here it is safer not to struggle for a perfect radiographic reduction.

The scaphoid serves as an important link between the proximal and distal rows of carpal bones, and without this link, degenerative arthritis gradually develops from abnormal carpal bone motions. Since the scaphoid fractures occur in young adults, anatomic reduction and healing of scaphoid fractures are important, not so much for short-term function, but to preclude degenerative arthritis 10 – 20 years later. The scaphoid is covered on nearly all surfaces by cartilage, its nutrient arteries are sparse, and the blood supply to the proximal pole may be disrupted following fracture. 95% of scaphoid fractures will eventually heal, but prolonged immobilization may be required because of poor blood supply. Twelve to sixteen weeks in a thumb spica cast may be required for healing. (Irrelevant fact: Spica means “ear of wheat” in Latin, and some poet saw the resemblance of the overlapping layers in a spica cast to a head of wheat.)

Distal radius fractures in adults disturb the normal alignment of the hand on the forearm, a deformity likened to the curve of a fork. (It’s called the “silver fork deformity”, although steel and plastic forks are shaped identically.) Not only is the deformity unsightly, but also wrist flexion and consequently a large portion of hand function are lost. Thus anatomic reduction and healing are sought, even more so when the fracture is comminuted with irregularity of the articular surface of the distal radius. This goal is frequently achieved by closed manipulation and splinting followed in several days by casting after local swelling has subsided. Premature placement of circumferential plaster risks vascular compromise from associated soft tissue swelling. At times, percutaneous pinning, use of an external fixator, or open reduction with internal fixation may be required to achieve and maintain the desired reduction. Complications following Colles’ fractures abound and include malunion, restricted forearm, wrist and finger joint motion, median nerve compression, reflex sympathetic dystrophy, and loss of forearm rotation due to disruption of the distal radio-ulnar joint. Despite the layman’s impression that “it’s just a wrist fracture”, most persons with Colles’ fractures are left with some residual dysfunction.

Questions:
1. Describe a common complication of fracture of the distal radius. (Several possibilities)
2. Persistent tenderness at a distal radius fracture site 4 – 6 weeks after the injury probably means that……..?
3. Define a non-union (of a fracture).
Objectives:
Through a series of case discussions, goals are: 1) to understand the most common clinical presentations and management of common pediatric lower extremity issues, such as septic hip, slipped capital femoral epiphyses, Perthes’ disease (AVN), and causes of intoeing in gait. 2) To understand what processes most commonly present at certain age/gender groups.


CASE #1
You are asked to see a 10-year-old male because of a five day history of anterior thigh, medial knee pain of his right lower extremity. He is an active lad but now has an antalgic gait. He has tried to increase his activity level recently because of parental concern of obesity.

1. At this point, what is your differential diagnosis?

2. What further history should you obtain?

On physical exam, the child walks with the right lower extremity in mild external rotation. He is afebrile. Knee exam is negative. There is limitation of gentle passive motion, specifically internal rotation.

3. How would you investigate further?

4. If an xray were taken of the hip and the entire femur, what would you expect to find in each of the above differential diagnoses?

5. What is your plan of management?
CASE #2
A 7-year-old female is brought to your office because of an in-toeing gait.

6. What questions relative to history should be asked?

7. How would you do the physical examination?

8. How would you decide the etiology of the in-toeing?

9. What would your management be?
Radiographs:

Perthes’ disease: AVN, femoral head collapse

SCFE: Left hip

Bilateral pinning

DDH: disrupted Shenton’s line

DDH: late changes – untreated.

Images courtesy of ImagesMD website.
Developmental Dysplasia of the Hip:

Incidence:
- Occurs in the neonatal period, therefore, not a true congenital issue.
- Hips are initially “dislocatable”, but progress to being eventually dislocated.
- Subtypes:  
  a. Typical: no neurological impairment as cause.
  b. Neurogenic: e.g. CP, spinal cord injury, muscular dystrophy.
- Affects an estimated 1: 100 newborns; 1: 1000 true dislocation.

Risk Factors:
- Multifactorial etiology – physiological and mechanical factors.
- Risk factors: “the five F’s”
  o Family: 20% have positive family history.
  o Flexible: General ligamentous laxity often seen, worse with maternal estrogens.
  o First born -- ~ 60% of those with DDH are #1.
  o Female: -- 9:1 female: male ratio.
  o Foot-first: ~ 30 – 50% developed in the breech position, especially Frank breech, which increases extreme hip flexion, and ligamentous stretch.
  o Associations: congenital torticollis: (~15%); metatarsus adductus (~5%)
    ▪ If either present, look for hip problems.
  o Later: acetabular maldevelopment, excessive femoral anteversion, and hip muscle contractures occur.

Clinical Assessment:
- Tests:
  o Barlow: “back out” -- try to dislocate an unstable hip.
    ▪ Stabilize pelvis, flex hips to 90°. Index finger on greater trochanter. Flex and adduct one hip at a time, applying a posterior force.
  
  o Ortolani: “in” -- -- try to reduce a dislocatable hip.
    ▪ Same as Barlow positioning, then Flex and Abduct the hip, with lifting femoral head anteriorly into the acetabulum.
    ▪ “Clunk” -- Not clicks, which can be normal.
    ▪ More common if > 1 month old.
  
  o Galeazzi: older infants
    ▪ Flex hips, knees to 90°, place feet on table, then observe knee heights – if one is out, then it will be shortened, with asymmetric # of thigh skin folds.
  
  - Radiography:
    ▪ Dynamic ultrasound – investigation of choice*.
    ▪ X-rays – Shentons’ line disruption.
Treatment:
- **Age-dependent:**
  - **Birth:**
    - Maintain in flexed, abducted position x 1-2 mo.
    - Abduction orthoses: Pavlik harness.
    - Double-diapering.
    - Rx until hips clinically stable.
  - **1 – 6 months:**
    - Pavlik harness (brace in wide abduction).
      - ~ 90-95% success if dislocatable; ~80% in dislocated.
    - If fails, surgical closed reduction with skin traction, adductor tenotomy with closed reduction, and hip spica casting after.
  - **> 6 months:**
    - Surgery:
      - Closed or open reductions.
      - Pelvis and/or femoral osteotomy, with 6/52 hip spica casting (in abducted position).
  - Complications: AVN of hip; sepsis.

**NOTE:** Clinical Examination with baby hip models will be offered informally if desired **AFTER** MS084 session. Please keep this part of the notes for that time.
Objectives:
MS044:
- To understand the nomenclature relating to neoplastic disease in bone
- To learn algorithms that are applied to the clinical investigation of patients who are suspected to have a neoplastic bone disease
- To become aware of the principles on which diagnosis of these diseases are based

MS045:
- To examine examples of common bone tumors and discuss issues involved in their diagnosis
- To discuss how errors can occur in tumor diagnosis

References: Ess Surg Specialities 3rd ed. Pg. 279 – 283, e.g. Table 6-9, pg. 282.

Notes:

1. Q: How are bone tumours diagnosed?
   A: Most bone tumours are diagnosed on the basis of local symptoms that they produce, such as pain due to compression of nerves or swelling of the extremities. Further workup of the patient is done by diagnostic team, which includes an orthopedic surgeon, a radiologist and a pathologist. Accordingly, all bone tumours are diagnosed only by correlating the following:
   1. Clinical data, such as age, sex of the patient, family history, previous diseases and location of the tumor
   2. X-rays findings, indicating whether the tumor is most likely benign or malignant
   3. Histopathologic data, essential in diagnosing the tumor as benign or malignant, and further classifying the tumor as bone forming, cartilagenous or other.

2. Q: Name the data most important for the diagnosis of bone tumours.
   A: Age: Most bone tumors show an age-dependent occurrence. For example, osteosarcomas and Ewing sarcomas are tumors of childhood and adolescence, whereas chondrosarcomas usually occur after 40 yr of age
   Anatomic site: Osteosarcomas occur predominantly in the long bones of the extremities, whereas chondrosarcomas tend to involve the axial skeleton of the body
   Part of the bone involved in the tumor: Osteosarcomas tend to occur in the metaphysis of long bones, whereas Ewing’s sarcoma involves the diaphysis
3. Q: What are the principles of bone tumour classification?

A: According to their cell of origin and histological features:

- Bone forming (e.g. osteoid osteoma, osteoblastoma, osteosarcoma)
- Cartilage forming (e.g. chordoma, chondrosarcoma)
- Osteoclastic cells (e.g. giant cell tumor)
- Fibroblastic cells (e.g. fibrosarcoma, malignant fibrous histiocytoma)
- Undifferentiated cells (e.g. Ewing sarcoma)
- Hematopoietic and lymphoid cells (e.g. multiple myeloma, leukemia, lymphoma)

4. Q: Which tumours occur most often in the epiphysis of long bones?

A: Giant cell tumors

5. Q: Which tumours occur most often in the metaphysis of long bones?

A: Osteosarcoma is most common malignant; non-ossifying fibroma is the most common benign tumor in this location

6. Q: Which tumours occur most often in the diaphysis of long bones?

A: Ewing sarcoma or metastasis from other primary site

7. Q: Which tumours occur most often in the short bones of the hands and feet?

A: Enchondromas; if multiple may undergo malignant transformation

8. Q: What is the most common benign tumor of bones?

A: Osteochondroma (exostosis)

9. Q: Name the main risk factors for osteosarcoma (OS)

A: most OS develop in persons who have no known predisposing conditions. In a minority of cases (referred as secondary OS) there are identifiable risk factors such as:

1. Retinoblastoma (RB-1) gene mutation or deletion -
2. Radiation therapy or exposure to radioactive isotopes
3. Paget disease

10. Q: Which other “small blue tumor” should be considered in the differential diagnosis of Ewing sarcoma (ES)?
A: **Neuroblastoma, malignant lymphoma, rhabdomyosarcoma** and **small cell carcinoma**; can be distinguish from ES by means of immunohistochemistry and chromosomal features

11. Q: Which tumors metastasize most frequently to the bones?

A: **Carcinomas of prostate, breast, lung, kidney, thyroid** and **gastro-intestinal tract** for adults; **neuroblastoma** in children. Typically, sites are areas of hematopoiesis, such as spine, ribs, skull, pelvis, and long bone metaphyses.

12. Q: What treatment options are for local metastases?

A: After tumour specific treatment with wide excision, chemotherapy, XRT, etc., local radiation for palliation of pain could be done. Prophylactic fixation of lesions > 50% diameter of a bony cortex (depending on the person’s overall health) is suggested to minimize pathological fracturing.

13. Q: Radiographic appearances of metastatic lesions can be:

A: - **Purely lytic** (kidney, lung, colon, and melanoma)
- **Purely blastic** (prostate and breast carcinoma)
- **Mixed lytic and blastic** (most common appearance)

14. Q: Which malignant tumor presents radiologically with multiple punched-out lesions in adults?

A: **Multiple myeloma**, which is the most common primary tumour of bone overall. Serum and urine protein electrophoreses are positive, but bone scans generally are not.

15. Q: What radiographic findings suggest **benign versus malignant** tumours?

A: (Anatomic site and part of the bone involved by the tumour, Number, Pattern of growth, Periosteal reaction, Pattern of matrix mineralization)
- **Location**: cortex or medulla; epiphysis, metaphysis, or diaphysis
- **Multifocal**: Both benign and malignant can be multifocal; benign: symmetrical
- **Pattern of growth**: Well-demarcated margin, **rim of sclerotic bone**: benign. Cortical expansion or destruction: malignant ‘Moth-eaten’ permeative pattern with multiple radiolucencies: highly malignant.
- **Periosteal reaction**: focal cortical thickening = “buttress”: slow growing **Elevated periosteum** = “hair on end”: rapidly growing None: benign more likely; metastases also possible
- **Matrix mineralization**: cloud-like amorphous densities: osteosarcoma Focal stippled densities, rings of calcification:
- Cartilage tissue (enchondroma, chondrosarcoma)
References: See notes.

ARTHRITIS IN CHILDHOOD

Objectives: At the completion of the lecture, the student will be acquainted with:
1) The clinical presentations of childhood rheumatic diseases and differential diagnosis;
2) The value and limitations of laboratory investigations;
3) Approach to therapy.

I. Classification of chronic arthritis in children (Table 1) – The diagnosis of chronic arthritis in children or adolescents requires:
- the presence of arthritis for at least 6 weeks;
- age at onset <16 years; and
- absence of other diagnoses.

Arthritis is defined as
- swelling of a joint or
- limitation of movement with joint pain or tenderness.

As a group chronic arthritis in children and adolescents is referred to as Juvenile Idiopathic Arthritis and is classified into 7 categories by the International League Against Rheumatism (ILAR) criteria as follows: (see also Tables 1 and 3).

1) Systemic arthritis – Definition: arthritis with or preceded by daily fever of at least 2 weeks’ duration accompanied by one or more of:
- evanescent rash
- generalised lymph node enlargement
- hepatomegaly or splenomegaly
- serositis
Fevers of up to 40°C occur in the late afternoon and evening with a return to normal or subnormal temperatures in the morning. The evanescen macular salmon-pink rash (“systemic rash”) often accompanies the fever. Pericarditis may occur.

2) Oligoarthritis (previously pauciarticular) - Definition: arthritis in ≤ 4 joint during the first 6 months. Usually large joints are affected. Most patients maintain an oligoarticular disease course (called persistent oligoarticular) but in about 20% more than 4 joints may be affected after the first 6 months (called extended oligoarticular). A complication is chronic asymptomatic uveitis, which may lead to blindness if untreated. There is a strong association between antinuclear antibody and young age of onset, female sex, and uveitis.

3) Polyarthritis rheumatoid factor negative – Definition: arthritis affecting ≥ 5 joints during the first 6 months of disease; negative rheumatoid factor. Disease course is polyarticular. Affects both large and small joints.

4) Polyarthritis rheumatoid factor positive – Definition: arthritis affecting ≥ 5 joints during the first 6 months of disease; positive rheumatoid factor. Onset is in late childhood or adolescence. Disease course is polyarticular. This type has a poor prognosis; arthritis is unremitting and there is a high risk for disability.
Systemic, oligoarthritis (pauciarticular), polyarthritis rheumatoid factor negative, and polyarthritis rheumatoid factor positive were previously called juvenile rheumatoid arthritis.

5) **Enthesitis-related arthritis** – (spondyloarthropathy): Definition:

**Arthritis and enthesitis or**

**Arthritis or enthesitis with 2 of:**

- sacroiliac joint tenderness and/or inflammatory spinal pain
- Histocompatibility antigen (HLA)-B27
- Family history of spondyloarthropathy in first degree relative
- Acute uveitis
- Male with onset of arthritis >6 years of age

This category is a juvenile form of ankylosing spondylitis. It forms a continuum and occurs principally in boys. Onset is usually after age 6 years. The large joints of the lower extremities are predominantly affected. Unlike in adults, low back pain is infrequent at onset. Arthritis and/or enthesitis are the main findings at onset and early in disease. Later low back pain due to sacroiliitis develops in about 25% of cases and the disease may progress to typical adult type ankylosing spondylitis. Once X-ray changes of sacroiliitis develop, the diagnosis can be changed to juvenile ankylosing spondylitis. As in adults, there is a strong association of spondyloarthropathy with HLA-B27.

Enthesitis is an inflammation at the insertion of tendons into bone. Common sites are the plantar and Achilles tendon insertions into the calcaneus resulting in pain of the heel.

6) **Psoriatic arthritis:**

**Definition:** Arthritis and psoriasis or

**Arthritis with at least 2 of:**

- Dactylitis
- Nail pitting or onycholysis
- Family history of psoriasis in a first degree relative

Psoriatic arthritis is slightly more common in girls than boys. Dactylitis (a diffuse swelling of the digits due to inflammation of the tendon sheath), distal inter-phalangeal joint involvement and nail pitting are characteristic. Psoriasis may precede (40% of cases) or follow (50%) or start simultaneously with arthritis (10%). Antinuclear antibody may be positive but rheumatoid factor is negative. Asymptomatic chronic uveitis may occur.

7) **Undifferentiated:** Patients not fitting any of the above categories or fulfilling criteria for more than one are put into this group.

The estimated incidence and prevalence of juvenile idiopathic arthritis in children in North America average 7/100,000 and 50/100,000, respectively.

**Other:** Although the conditions below are part of the spondyloarthropathy group, they are not included in the Juvenile Idiopathic Arthritis classification:

Arthritis with inflammatory bowel disease (Crohn’s disease or ulcerative colitis): Two types of arthropathy occur. One consists of arthritis of peripheral joints and occurs when the bowel disease is active. The other type affects the spine and sacroiliac joints, has no relation to activity of the bowel disease, and is associated with HLA-B27.
Reactive arthritis develops after genitourinary or gastrointestinal infection. This is rare in the pediatric age group. It is a spondyloarthropathy, but because it has a known etiology, it is not included under juvenile idiopathic arthritis. Reactive arthritis used to be called Reiter’s syndrome if the triad of arthritis, conjunctivitis, and urethritis are present, but the latter name has been dropped.

II. Investigations: Diagnostic “arthritis tests” do not exist. Rather, a clinical diagnosis of chronic arthritis should be made first. The erythrocyte sedimentation rate (ESR) may be elevated during active disease but elevations may be minimal or absent in oligoarticular arthritis. Antinuclear antibody and rheumatoid factor are not diagnostic, and have poor specificity and poor sensitivity for juvenile idiopathic arthritis. The risk of uveitis is increased if the antinuclear antibody is positive; rheumatoid factor is used to classify patients with polyarticular arthritis (see above and Table 3).

X-rays: Early changes are soft tissue swelling and periarticular osteoporosis. If arthritis is asymmetric, advanced development of the epiphysis on the affected side is often seen. Erosions and joint space narrowing take time to develop.

III. Treatment of chronic arthritis.
   a) Medications:
      Nonsteroidal anti-inflammatory drugs (NSAID’s).
      Disease modifying anti-rheumatic drugs (DMARD): methotrexate, sulphasalazine, hydroxychloroquine, gold salts. Most commonly methotrexate is added for patients with rheumatoid factor negative or rheumatoid factor positive polyarthritis. For patients with rheumatoid factor positive polyarthritis, DMARD’s should be started early.
      Corticosteroids: intraarticular steroids especially for limited joint involvement; systemic corticosteroids for control of fevers or pericarditis in systemic arthritis.
      Biologic therapies: These agents specifically antagonize one or more steps in the immune response leading to inflammation. The more established agents antagonise the effect of inflammatory cytokines, for example tumour necrosis factor-α inhibitors; more recent products interfere with cell to cell interaction. Biologic therapies are reserved for patients with resistant disease.

b) Physical and Occupational Therapy: aimed at maintaining joint function.

c) Ophthalmologic - Regular slit lamp examinations are required for the detection of asymptomatic uveitis. Treatment of acute or chronic uveitis includes topical steroids and mydriatics.

IV. Acute rheumatic fever: This is a post infectious condition occurring after pharyngeal infection with Group A streptococcus. The arthritis is usually migratory, lasting 3 to 6 weeks altogether if untreated. It is distinguishable from chronic arthritis by exquisite tenderness and pain of affected joints and the prompt relief of pain with nonsteroidal anti-inflammatory drugs. Commonly, symptoms in one joint wane as those in another wax. Hence the term migratory. Evidence of a recent streptococcal infection is essential for the diagnosis. See Table 2.

V. Connective tissue diseases (Table 1).
   a) Systemic Lupus Erythematosus (SLE): SLE in children and adults is similar and is described elsewhere.
   b) Scleroderma - except for localized scleroderma (morphea and linear scleroderma), scleroderma is rare in childhood.

c) Dermatomyositis: Dermatomyositis generally occurs between 5-15 years of age (average 7 yrs.) with a male to female ratio of 1:2. It presents as muscle pain, weakness, rash + fever. The proximal muscles are most severely affected and frequently they are tender to touch. The rash consists of a heliotrope discoloration of the periorbital...
area, a malar rash, and bright pink-red atrophic changes (Gottron's papules) over the metacarpophalangeal and interphalangeal joints, elbows, and knees. The combination of symmetric proximal muscle weakness and typical rash is pathognomonic of dermatomyositis.

**Laboratory features:** Muscle enzymes (CPK, LDH, and SGOT) are elevated; electromyography shows denervation potentials, myopathic units and fibrillations. Muscle biopsy shows infiltration with lymphocytes, degenerating and regenerating fibres and necrosis. Diagnosis can be made on the basis of typical rash, weakness, and elevation of muscle enzymes. If any of these are missing, it is wise to confirm the diagnosis by ultrasound, magnetic resonance imaging, electromyography or biopsy.

**Treatment:** Corticosteroids, methotrexate, and intravenous immunoglobulin are all used. Physical therapy is required to maintain range of motion, correct contractures, and strengthen muscles.

Complications and residua of the disease include gastrointestinal hemorrhage due to vasculitis, joint contractures, muscle atrophy and calcinosis. Calcinosis occurs after the acute phase in about 40% of patients. Dermatomyositis is a unicyclic disease in nearly half of cases. The remainder have a recurrent or chronic course. Unlike the disease in adults, there is no association with malignancy.

**VI. Vasculitis (Table 1)**. Henoch Schonlein Purpura and Kawasaki Disease are the most common vasculitides in children.

a) A diagnosis of Henoch Schonlein Purpura, a small vessel vasculitis, is based on the presence of abdominal pain, arthritis and purpura. Renal involvement occurs in 20 to 30%.

b) Kawasaki disease, a small and medium vessel vasculitis, affects young children, most commonly under 2 years. The male to female ratio is 1.5:1. Diagnostic criteria are fever for >= 5 days without other cause and at least 4 of the following criteria: rash, conjunctivitis, mucocutaneous changes (reddening of the lips, tongue, oral mucosa), cervical lymph node enlargement, and changes in the extremities (swelling of the hands and feet, erythema of palms and soles, and desquamation of fingers and palms, toes and soles). Other signs and symptoms occurring less commonly are arthritis, aseptic meningitis, sterile pyuria, myocarditis and pericarditis. Major complications relate to coronary aneurysms and thrombosis. Treatment consists of aspirin and intravenous immunoglobulin to reduce the risk of coronary artery complications.

**VII. Differential diagnosis of joint swelling**

**Infectious arthritis:**

- **Bacterial**: The onset of septic arthritis is acute and includes fever, joint pain and swelling involving a single, usually large joint. The affected joint is markedly tender and painful on any movement. A patient with bacterial osteomyelitis may have an associated septic arthritis.

- **Viral infections** which may be associated with transient arthritis include Parvovirus, Epstein Barr virus, hepatitis B, rubella, mumps, and Varicella.

- **Mycoplasma infections** may also be associated with a self-limited arthritis.

**Other conditions** which cause joint swelling are malignancy (especially leukaemia), and trauma.

Kiem Oen, MD
Updated August 11, 2008
### Table 1. Classification of rheumatic and connective tissue diseases in children

<table>
<thead>
<tr>
<th>I. Chronic arthritis</th>
<th>II. Vasculitis</th>
</tr>
</thead>
<tbody>
<tr>
<td>Juvenile idiopathic arthritis:</td>
<td></td>
</tr>
<tr>
<td>1) systemic arthritis</td>
<td>1) Henoch-Schönlein purpura</td>
</tr>
<tr>
<td>2) oligoarticular</td>
<td>2) Kawasaki disease</td>
</tr>
<tr>
<td>3) polyarthritis rheumatoid factor negative</td>
<td>3) Antineutrophil cytoplasmic antibody positive vasculitis</td>
</tr>
<tr>
<td>4) polyarthritis rheumatoid factor positive</td>
<td></td>
</tr>
<tr>
<td>5) enthesis-related arthritis</td>
<td>4) Polyarteritis nodosa and cutaneous polyarteritis</td>
</tr>
<tr>
<td>(spondyloarthropathy)</td>
<td>5) Takayasu arteritis</td>
</tr>
<tr>
<td>6) psoriatic arthritis</td>
<td></td>
</tr>
<tr>
<td>7) undifferentiated</td>
<td></td>
</tr>
</tbody>
</table>

### Table 2. Jones criteria for the diagnosis of acute rheumatic fever

| Major manifestations: Carditis, polyarthritis, chorea, erythema marginatum, subcutaneous nodules. |
| Minor manifestations: Fever, arthralgia, increased ESR or C-reactive protein, prolonged P-R interval. |

**Supporting evidence for streptococcal infection:** Throat culture positive for Group A streptococci; increased or rising ASOT or other streptococcal antibodies.

**Requirements for a diagnosis:** 2 major or 1 major and 2 minor manifestations, in addition to the presence of evidence of a recent streptococcal infection indicates a high probability of acute rheumatic fever.
Table 3. Chronic arthritis in children and adolescents – juvenile idiopathic arthritis

<table>
<thead>
<tr>
<th></th>
<th>Systemic arthritis</th>
<th>Oligoarticular (pauciarticular) arthritis</th>
<th>Polyarthritis, RF negative</th>
<th>Polyarthritis, RF positive</th>
<th>Enthesitis-related arthritis (spondyloarthropathy)</th>
<th>Psoriatic arthritis</th>
</tr>
</thead>
<tbody>
<tr>
<td>% of all children and adolescents with chronic arthritis</td>
<td>11%</td>
<td>44%</td>
<td>16%</td>
<td>8%</td>
<td>15%</td>
<td>5</td>
</tr>
<tr>
<td>Peak age of onset (yrs)</td>
<td>No peak</td>
<td>1-3</td>
<td>1-3 and 8-10</td>
<td>&gt;9</td>
<td>&gt;8</td>
<td>&lt;5 and around 10</td>
</tr>
<tr>
<td>F:M</td>
<td>1:1</td>
<td>3:1</td>
<td>3:1</td>
<td>3:1</td>
<td>1:7</td>
<td>1.5:1</td>
</tr>
<tr>
<td>Uveitis</td>
<td>Rare</td>
<td>Chronic, asymptomatic</td>
<td>Chronic asymptomatic</td>
<td>Rare</td>
<td>Acute symptomatic</td>
<td>Usually chronic asymptomatic</td>
</tr>
<tr>
<td>ANA positive</td>
<td>Rare</td>
<td>65-85%</td>
<td>50-60%</td>
<td>50-60%</td>
<td>Negative</td>
<td>30-60%</td>
</tr>
<tr>
<td>RF positive</td>
<td>Negative</td>
<td>Negative</td>
<td>Negative</td>
<td>100% positive</td>
<td>Negative</td>
<td>Negative</td>
</tr>
<tr>
<td>Course</td>
<td>Systemic symptoms may be prolonged but usually resolve; Arthritis may be unremitting</td>
<td>Good articular prognosis; uveitis may leave morbidity</td>
<td>May be unremitting</td>
<td>Generally unremitting</td>
<td>May progress to involve sacroiliac joints and spine</td>
<td>May be unremitting</td>
</tr>
</tbody>
</table>


Genetics of Musculoskeletal Disease
University of Manitoba
Faculty of Medicine
Med II/MS076
Dr. C. Greenberg
2008-2009

Objectives:
After the lecture the student will
1. Be able to name 3 types of genetic constitutional disorders of bone and provide an example of each.
2. Be able to explain how certain multisystem metabolic bone disorders (e.g., Lysosomal storage diseases) lead to bone pain and bone deformities.
3. Be able to identify the pattern of inheritance of the most common muscular dystrophies.
4. Appreciate that the clinical classification of disorders of bone is evolving in parallel with the molecular classification.

Overview:
1. Constitutional disorders of bone
2. Hereditary Metabolic Bone Disorders
3. Primary muscle disorders

This lecture will attempt to provide an overview of the genetic musculoskeletal disorders. This lecture will not discuss or review disorders of calcium, phosphorus and parathyroid hormone metabolism which were covered elsewhere.

1. Constitutional disorders of bone

Genetic disorder of the skeleton are a large group of disorders which share certain features in common—specifically malformation, disproportionate growth and deformation of the skeleton as a whole or individual bones or groups of bones.

Currently they are best diagnosed by findings in 3 equally important categories.
A) Clinical features B) Radiographic analysis and C) Molecular findings

The 3 main groups in the constitutional disorders of bone include
1. Skeletal dysplasias or osteochondrodysplasias
2. Dysostoses

There are now ~ 33 subgroups within category 1 and 3 groups of genetically determined dysostoses. Skeletal dysplasias are generalized developmental disorder of chondro-osseous tissue.

Dysostoses are localized static skeletal malformations that occur singly or in combination. The dysostoses occur early in embryonic life whereas skeletal dysplasias often appear later, involve the skeleton as a whole or evolve throughout life.

Examples of different types of skeletal dysplasias will be presented as well as the dysostoses. Skeletal dysplasias can be lethal (thanatophoric dysplasia) or compatible with long life (achondroplasia). Many are
associated with disproportionate short stature (rhizomelic, mesomelic or acromelic shortening of bones) or with decreased bone density and bone fragility (the osteogenesis imperfectas). The best know group of dysostoses. Include those localized disorders with predominantly cranial and facial involvement = the craniosynostoses or predominantly axial involvement (the spondylocostal dysplasias).

2. **Hereditary Metabolic Bone Disorders**

Excluding primary disorders of calcium, phosphorus sand vitamin D, this group of disorders id highlighted by multisystem inborn errors of metabolism involving deficiencies of lysosomal enzymes. Accumulation of precursor substances that are stored within the lysosomes of macrophages of the RE system including the bone marrow lead to bony disease. Gaucher disease will be used as a representative example. Advances in the treatment of metabolic bone diseases using enzyme replacement therapy or bone marrow or stem cell transplantation are one of the most exciting in this area of metabolic genetics.

3. **Primary Muscle Disorders**

The muscular dystrophies are hereditary disorders characterized by muscle wasting and weakness. They can be classified in many ways – congenital, childhood or adult onset, static or progressive, affected the CNS or sparing the CNS. The elucidation of the molecular basis for many of the muscular dystrophies has allowed the emergence of more than one pathogenetic mechanism leading to muscle cell death. Representative examples will be shown.

References in addition to standard genetic text

3. Select web sites
   - [www.mdausa.org](http://www.mdausa.org) Muscular Dystrophy
   - [www.oif.org](http://www.oif.org) Osteogenesis Imperfecta
   - [www.genzyme.org](http://www.genzyme.org) Lysosomal Storage Diseases
Objectives
1. Describe common injuries of the upper extremity in children
2. Describe a few congenital abnormalities of the upper extremity
3. Understand the approach to the child with a neuromuscular disorder and deformity of the upper extremity

Normal Development
Children are not just small adults
Need to consider growth and development

Important milestones for the hand: Grasp objects 4 months
Transfer objects 6 months
Pinch 12 months
Throw ball 18 months

Amount of growth: Proximal humerus 80%
Distal humerus 20%
Proximal radius 25%
Distal radius 75%
Proximal ulna 20%
Distal ulna 80%

Injuries to the Upper Extremity
In a longitudinal study from New Zealand, 51% of boys and 40% of girls will experience a single fracture by age 18. Wrist and forearm fractures are the most common and account for 24% of all fractures.

Participating in certain sports such as snowboarding, horseback riding and soccer (goalie) was noted to increase the risk of wrist fractures.

Snowboarders were found to have an increased risk of elbow dislocations.

Ligamentous laxity may increase the risk of certain fractures. Supracondylar fractures of the distal humerus are associated with hyperextension of the elbow.

Fractures in children can have several forms.
Plastic deformation bending of the bone
Buckle or torus failure of the cortex on the compression side of the bone – e.g. in distal radius, the dorsal cortex crumples but the volar cortex remains intact

Greenstick incomplete fracture of long bone with failure of the cortex on the tension side of the bone

Complete
If the fracture involves the physis, the Salter-Harris Classification is used.

One must be able to describe a fracture both clinically and on radiographs. Consider location of fracture, amount of angulation, amount of displacement, and the presence or absence of malrotation.

General principles: Individualize fracture treatment based on patient factors and the “personality of the fracture”
Consider amount of remaining growth
Consider activity of physis – a more active physis will remodel more completely and quickly
Damage to an active physis will cause more deformity

Two examples of pediatric upper extremity injuries:
Case 1 – Fracture of the distal radius
Case 2 – supracondylar fracture of the distal humerus

**Congenital Anomalies of the Upper Extremity**
- Congenital anomalies affect 1-2% of newborns
- Approximately 10% will involve the upper limb
- Certain anomalies occur in isolation, while others are part of a syndrome
- Classification: embryological, teratological or anatomical
- Most common classification scheme (used by ASSH)
  1. failure of formation
  2. failure of differentiation
  3. duplication
  4. overgrowth
  5. undergrowth
  6. constriction band syndrome
  7. generalized skeletal abnormalities
Incidence of the most common deformities
- Syndactyly: 18.2%
- Polydactyly: 14.6%
- Camptodactyly: 6.9%
- Amputations: 6.8%
- Clinodactyly: 5.5%
- Brachydactyly: 5.2%
- Radial club hand: 4.6%
- Central deficiencies: 3.9%

Evaluation of patient and family
- Address the shattered perception of the “perfect” child
- Adaptation
- Design a careful, individualized treatment plan for each child

Case 1: Radial hypoplasia (radial clubhand)
- Affects the preaxial border of the limb
- Ranges from mild thumb hypoplasia to complete absence of the radius
- *Found frequently with other anomalies*
- Most common syndromes: Fanconi’s anemia, TAR, Holt-Oram, VACTERL
- Need to consider the whole child – pay particular attention to heart, kidneys and platelet status

Case 2: Syndactyly
- Failure of the normal process of separation
- Failure of apoptosis (programmed cell death)
- Normal separation starts at the tip of the fingers and proceeds proximally
- 1:2000 live births
- Can be isolated or part of a syndrome
  - Simple – only skin and subcutaneous tissues
  - Complex – involves skeletal elements
  - Complete – webs extends to the fingertips
  - Incomplete – interdigital web is longer than normal, but not to tip
- Most common – involves the long/ring interspace
- Associated syndromes: Poland, Apert, amniotic disruption sequence

Case 3: Polydactyly
- Extra digit
• Preaxial, postaxial, or central
• Most common is postaxial polydactyly

Neuromuscular Disorders affecting the Upper Extremity
Case example: Cerebral Palsy

• Static, nonprogressive disturbance of the cerebral cortex occurring before the age of two
• Results in altered motor, sensory and often intellectual function
• Remember: while the brain lesion may be permanent and non-progressive, the natural history of cerebral palsy is not static
• Growth and maturation of the central nervous system, and the whole child will cause changing musculoskeletal problems
• In 1843 William J. Little first described “spastic rigidity of the limbs of newborn children” – initially called “Little’s disease”
• Most of the focus has been on the lower extremity
• Prevalence – 1 to 7 per 1000 live births
• Classification: neuropathic (ie spastic, athetoid, hypotonic, mixed) or anatomic (quadriplegic, diplegic, hemiplegic)
• Most common presentation to a hand surgeon: most parents notice a problem with the upper extremity by age 1 year
  o Lack of refined pinch (opposition of thumb to index)
  o Development of key pinch (thumb to side of index)
• Evaluation:
  o important to discuss expectations
  o will not achieve a “normal” limb
  o team approach
  o beware of the “evil grandmother”
• Physical examination
  o Voluntary hand use
  o Motor function
  o Sensibility
  o Intelligence/ability to comply
  o Presence of movement disorders
• Goals
  o Improved function
  o Improved appearance
  o Improved hygiene
• Timing of surgical intervention
• Delay surgery until a clear evaluation of functional use is possible – age 6-12 year
  • Surgery in older individuals can lead to functional improvement

• Surgical options
  • Weakening of spastic muscles by tendon/muscle transfer
  • Enhancing the antagonistic muscle function by augmentation tendon transfer
  • Permanently positioning a part by joint arthrodesis to provide best functional position