OB GYN SONOGRAPHY REVIEW

Fetal Skeleton

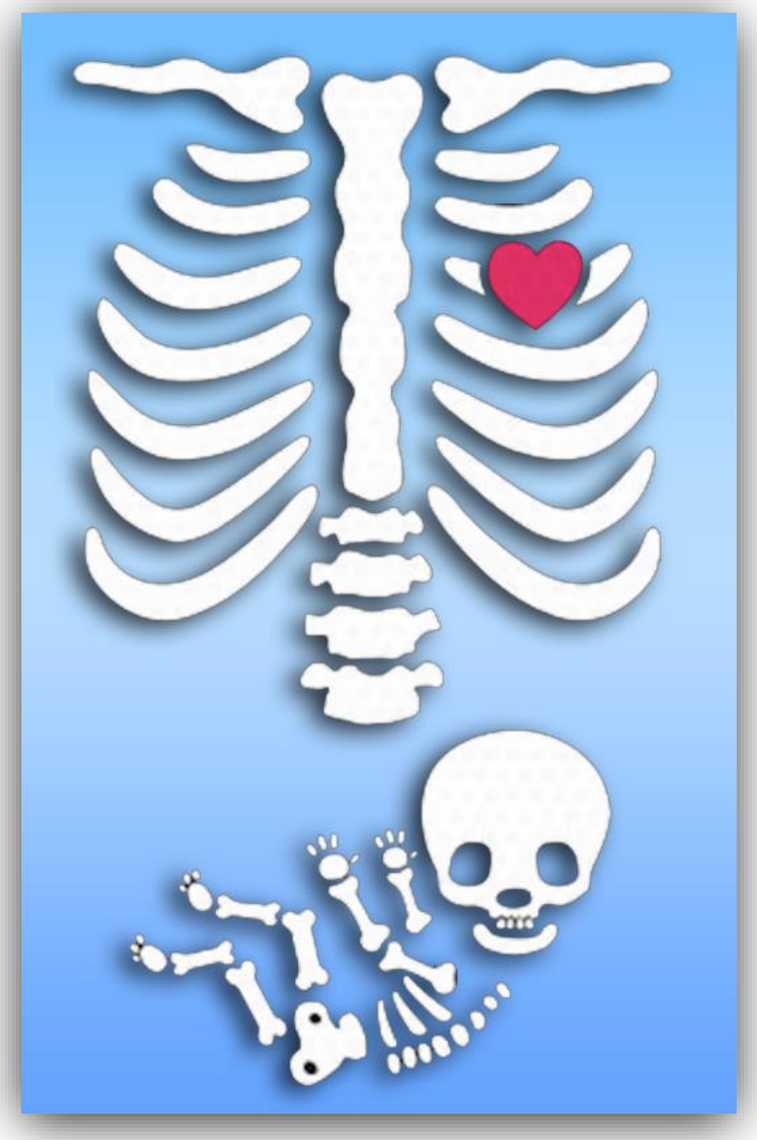
Course Outline

- Normal Sonographic Anatomy
  - Axial skeleton
  - Appendicular skeleton
- Skeletal Abnormalities

Normal Sonographic Anatomy

Axial Skeleton

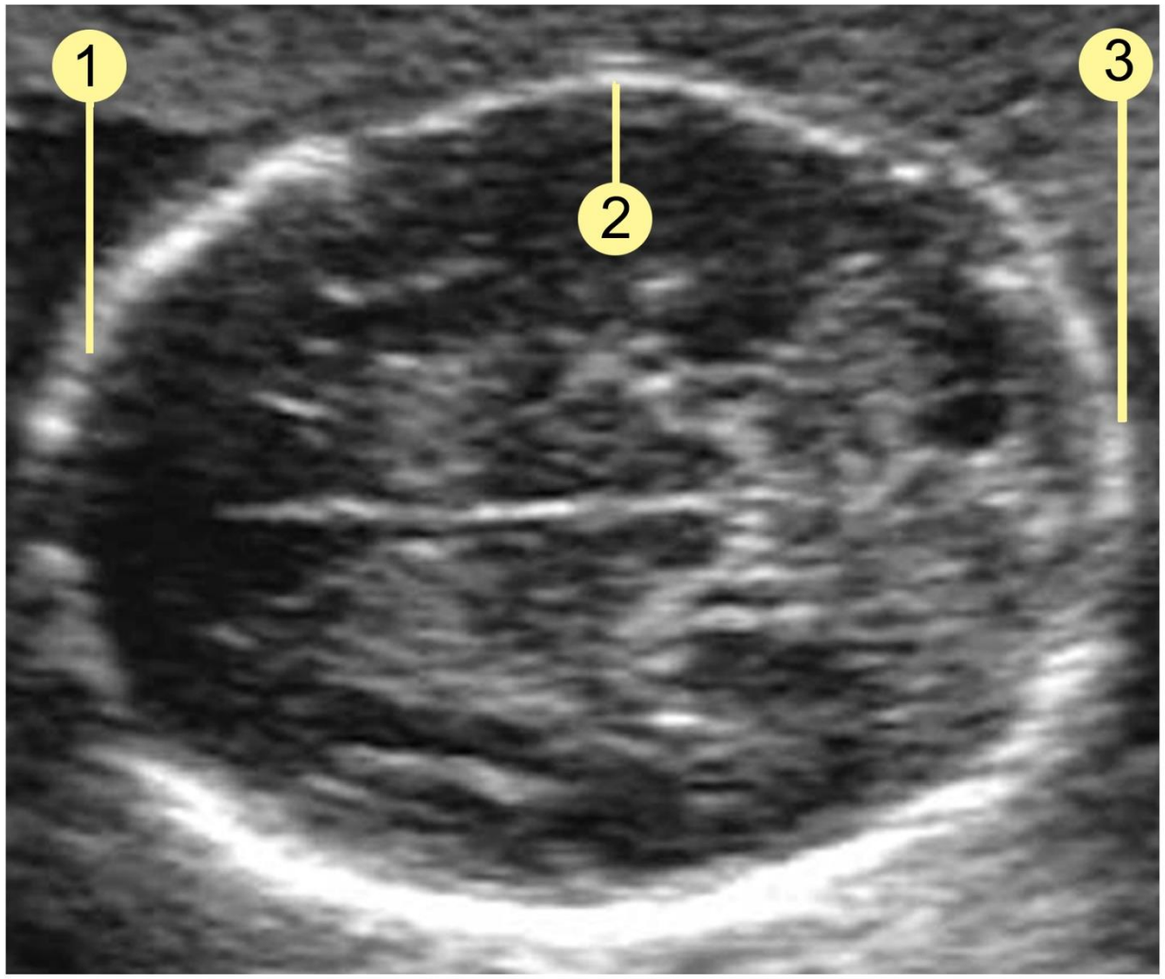
- The axial skeleton consists of:
  - Cranial bones
  - Facial bones
  - Pelvis
  - Spine



Cranial Bones

- Frontal, temporal, occipital, parietal bones
- Sphenoid bone & petrous ridges:
  - Separate cranial fossae: *anterior, middle, posterior*

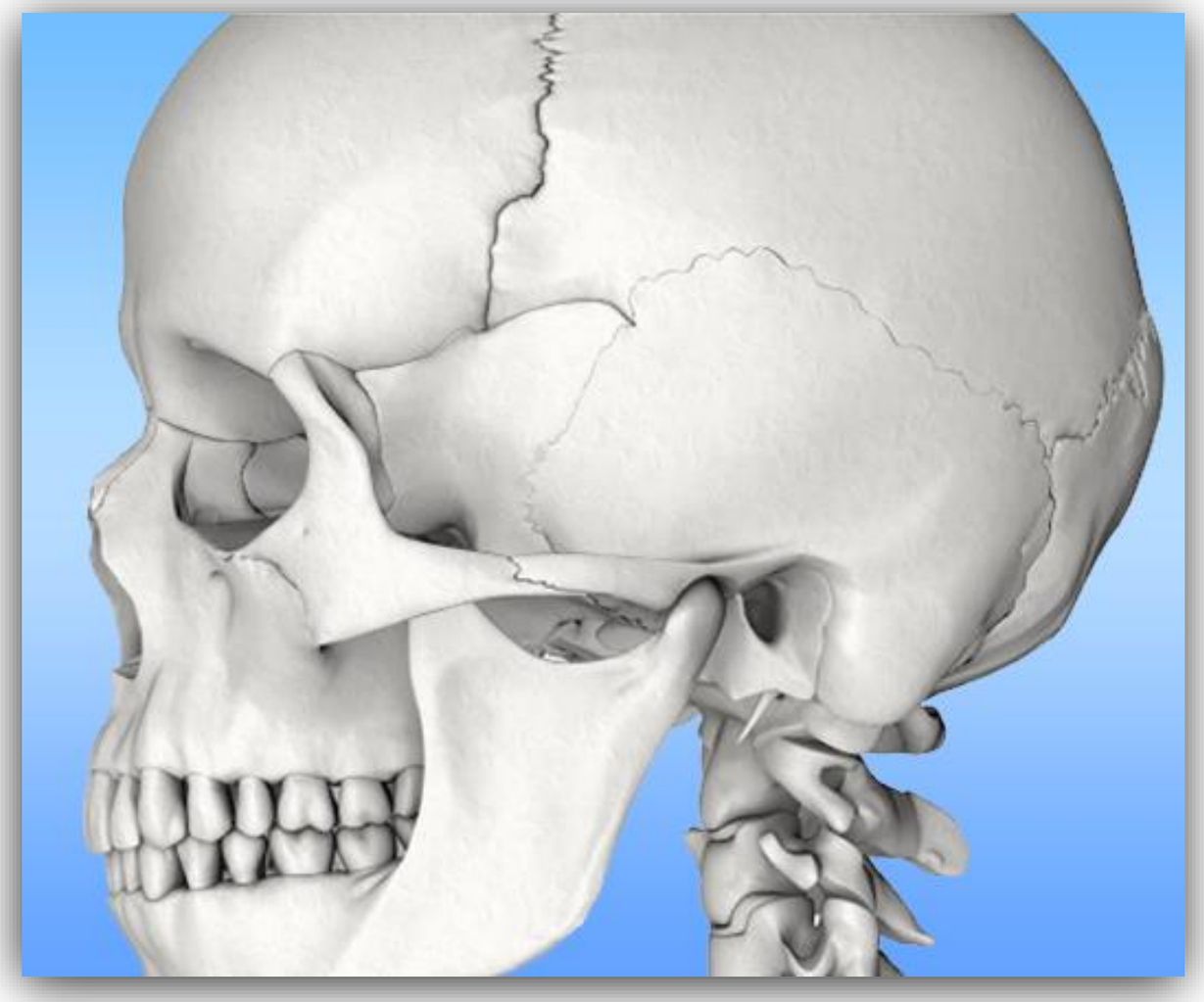
CRANIAL BONES



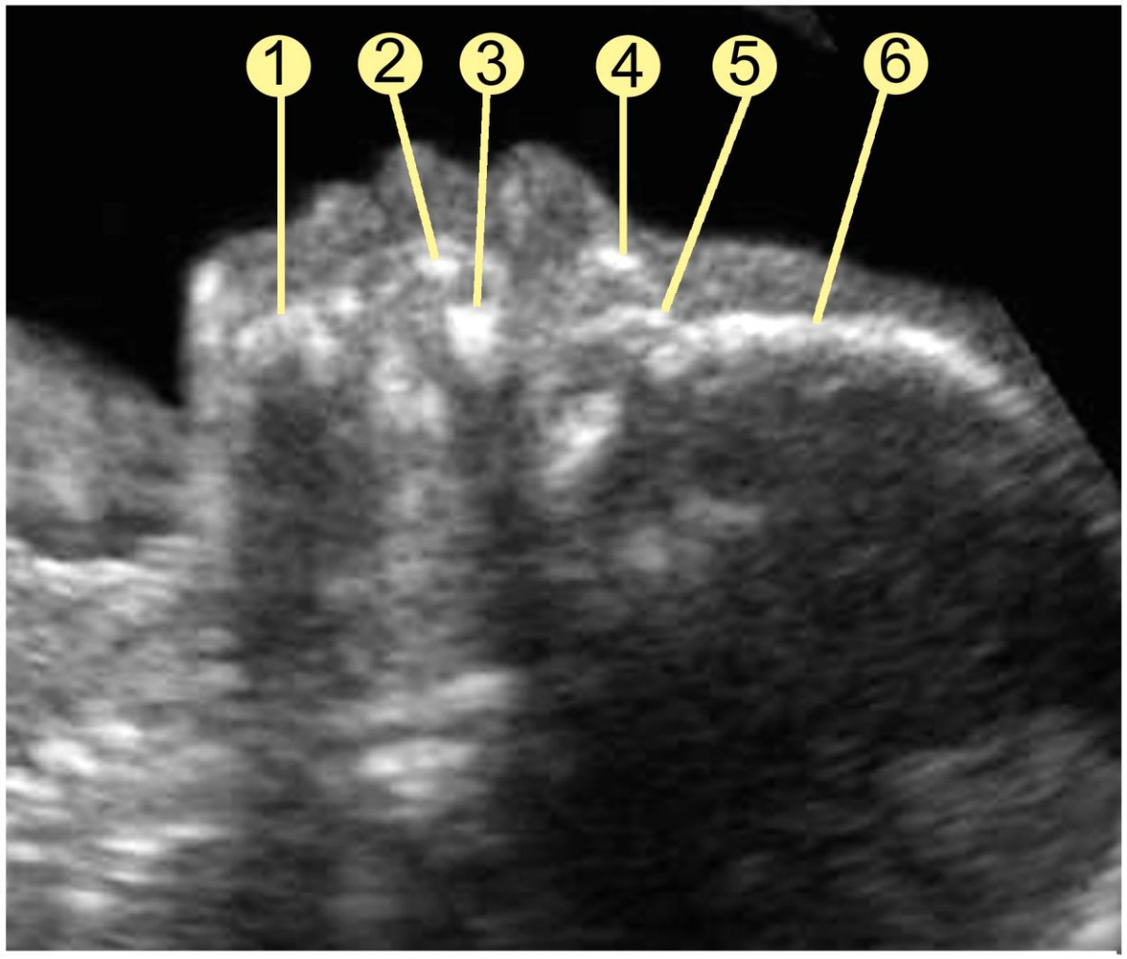
1 = frontal bone
2 = parietal bone
3 = occipital bone

Facial Bones

- Orbits
- Maxilla
- Mandible
- Bony nasal septum



FACIAL BONES

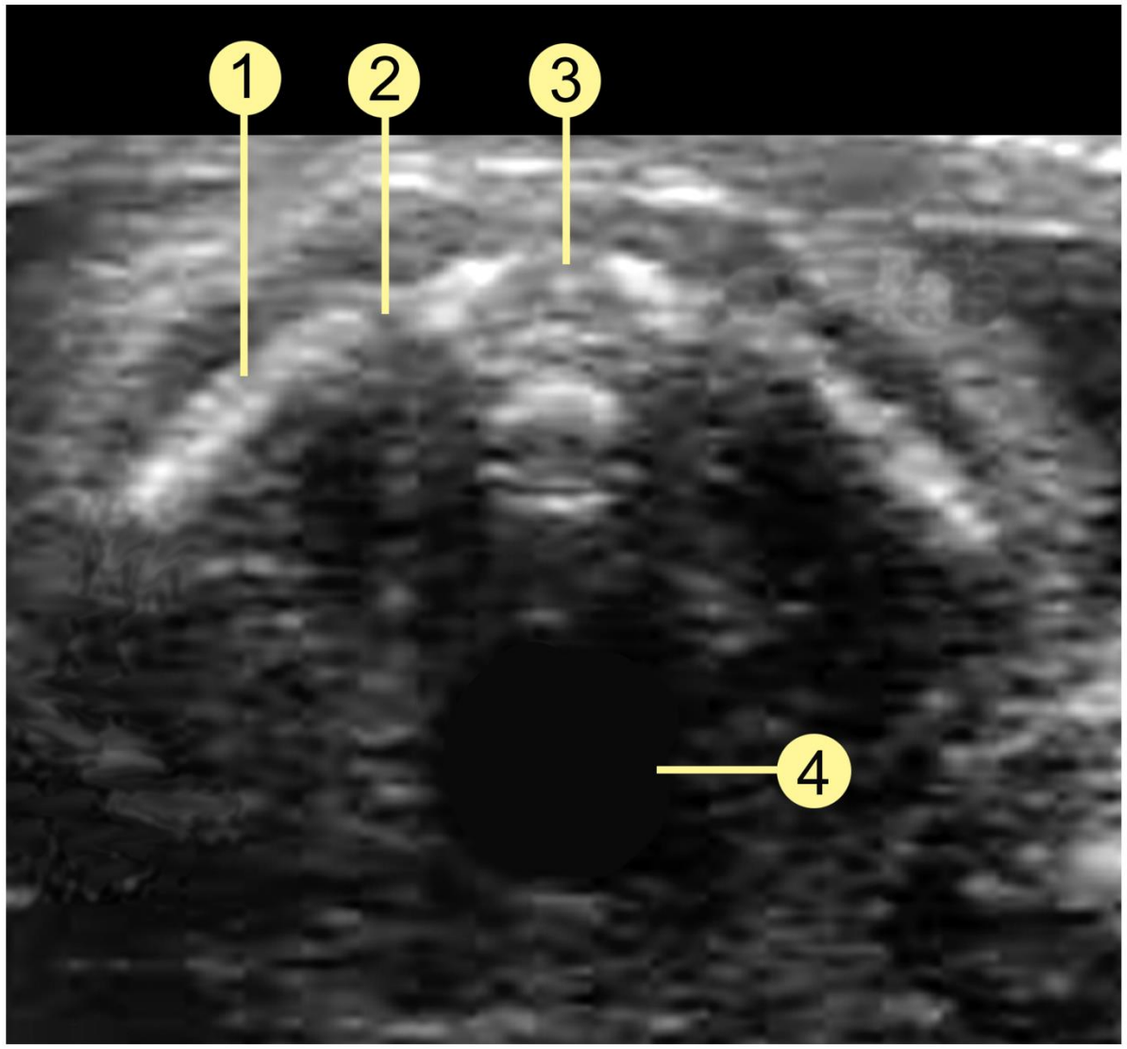


1 = mandible
2 = tooth bud
3 = maxilla
4 = nasal bone
5 = orbital rim
6 = frontal bone

Pelvic Bones

- Iliac ossification centers seen from early 2nd trimester
- Ischial ossification centers seen at ≈ 20 weeks
- Sacrum and sacroiliac joints

PELVIC BONES

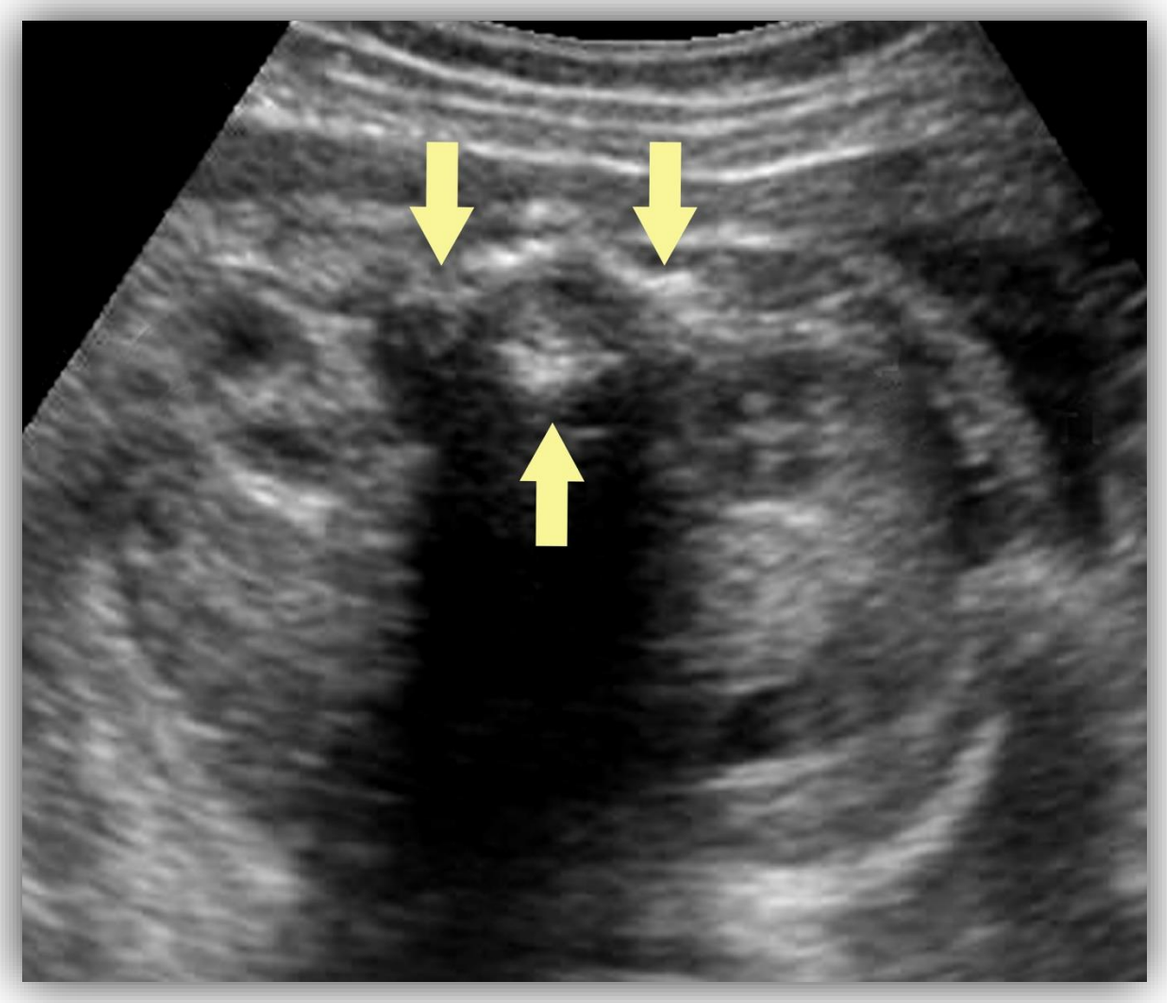


1 = iliac wings
2 = sacroiliac joint
3 = sacrum
4 = urinary bladder

Spine

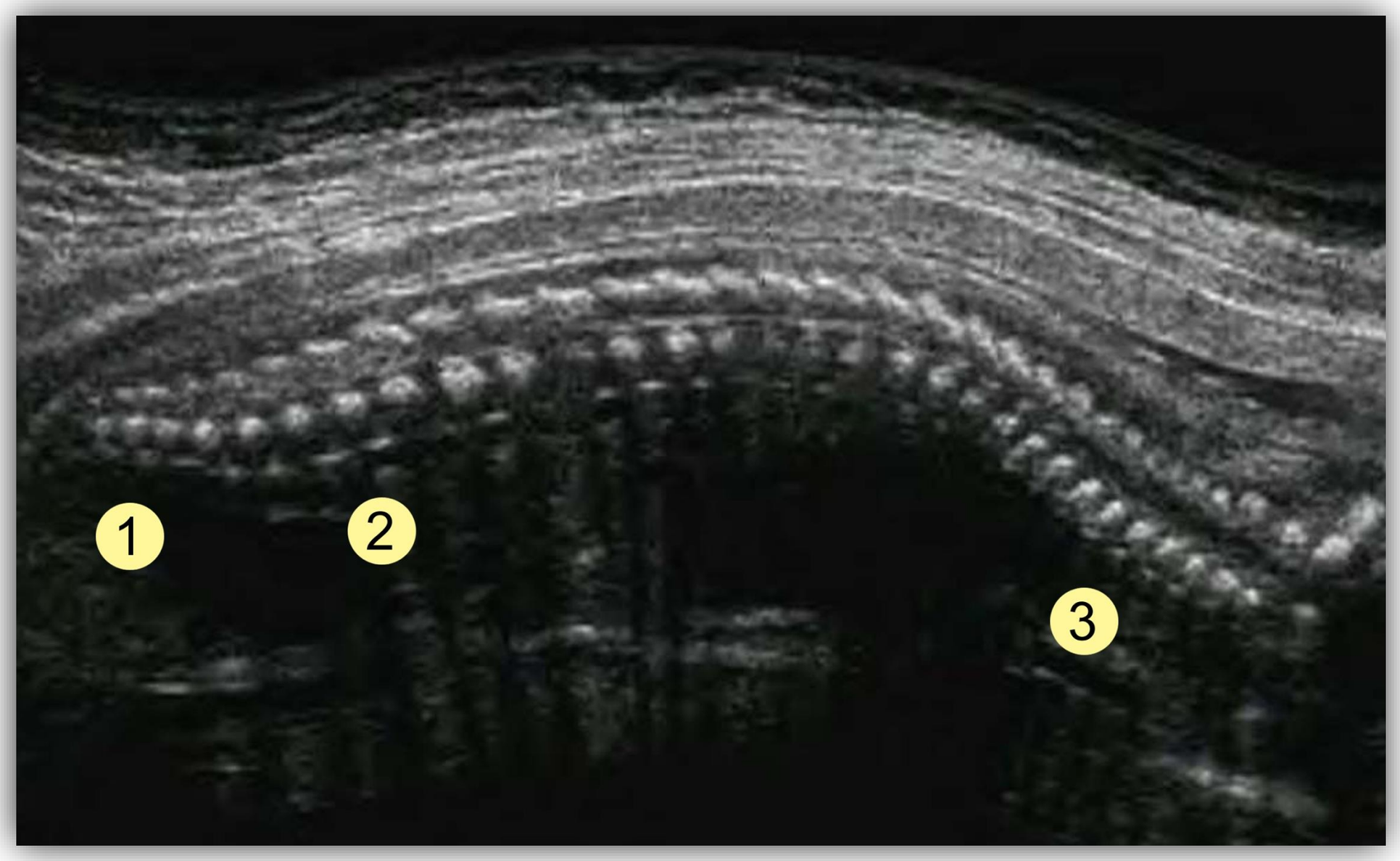
- Three ossification centers in each vertebra
  - 1 anterior (vertebral body)
  - 2 posterior (between lamina and pedicles)
- Spine can be visualized with great clarity after 22 weeks

SPINE



Location, configuration, number of ossification centers in each vertebra

NORMAL SONOGRAPHIC ANATOMY - SPINE

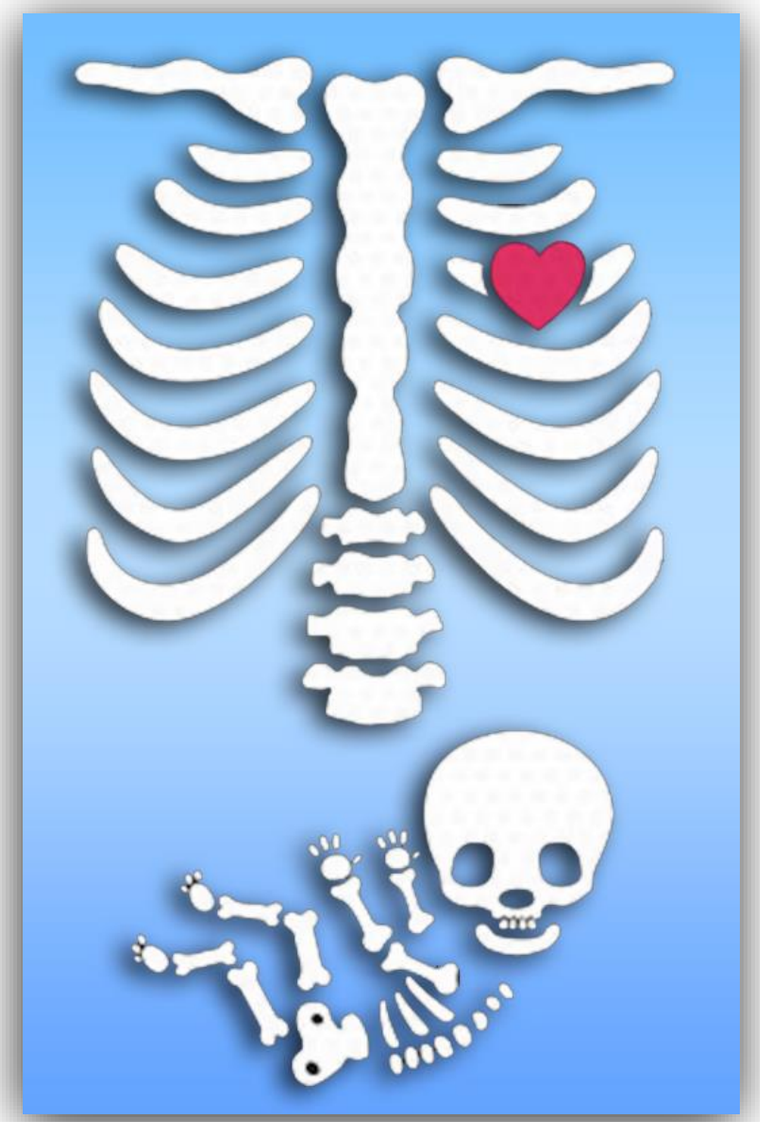


Sagittal spine

1 = sacral-caudal tapering
2 = lumbosacral curvature
3 = cervical curvature

Appendicular Skeleton

- The appendicular skeleton consists of:
  - Bones of the appendages
  - Upper extremity
  - Lower extremity



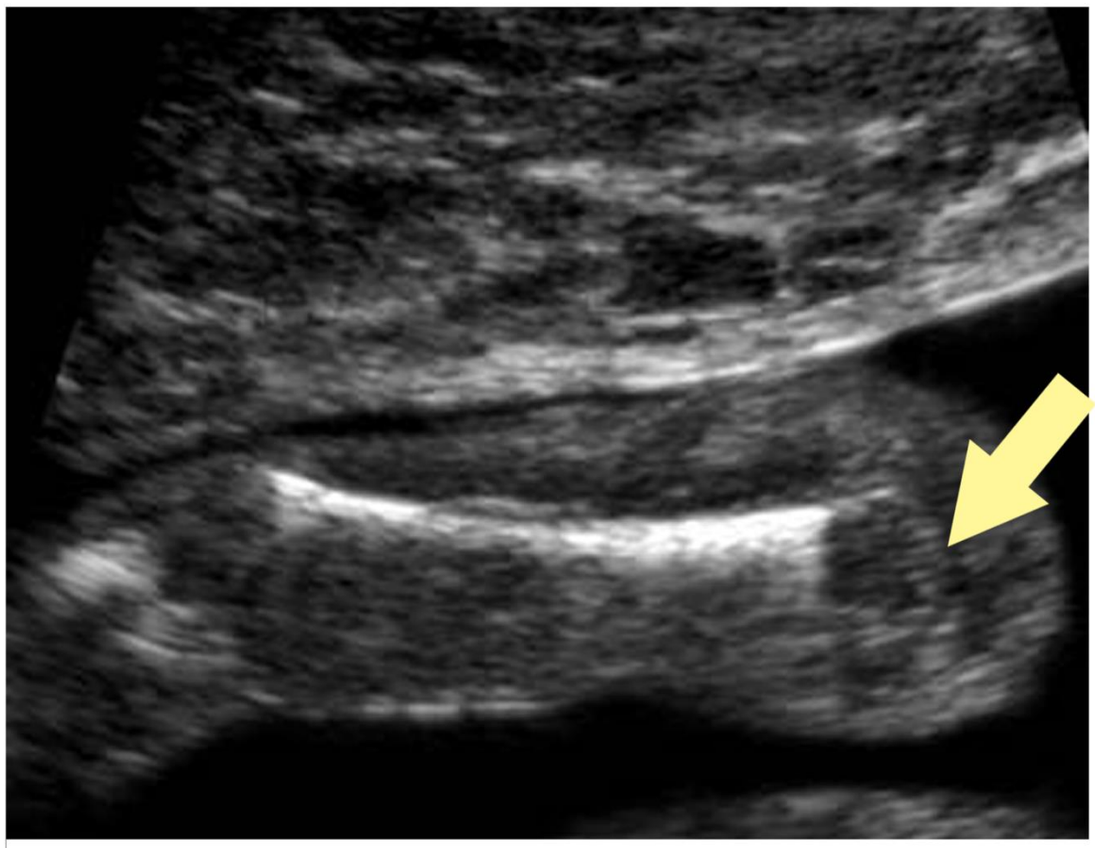
Appendicular Skeleton

- Upper extremity bones
  - Scapulae & clavicles ≈ 7 weeks
  - Metacarpals ≈ 16 weeks
  - Radius & ulna ≈ mid 2nd trimester
  - Humerus ≈ mid 2nd trimester

Appendicular Skeleton

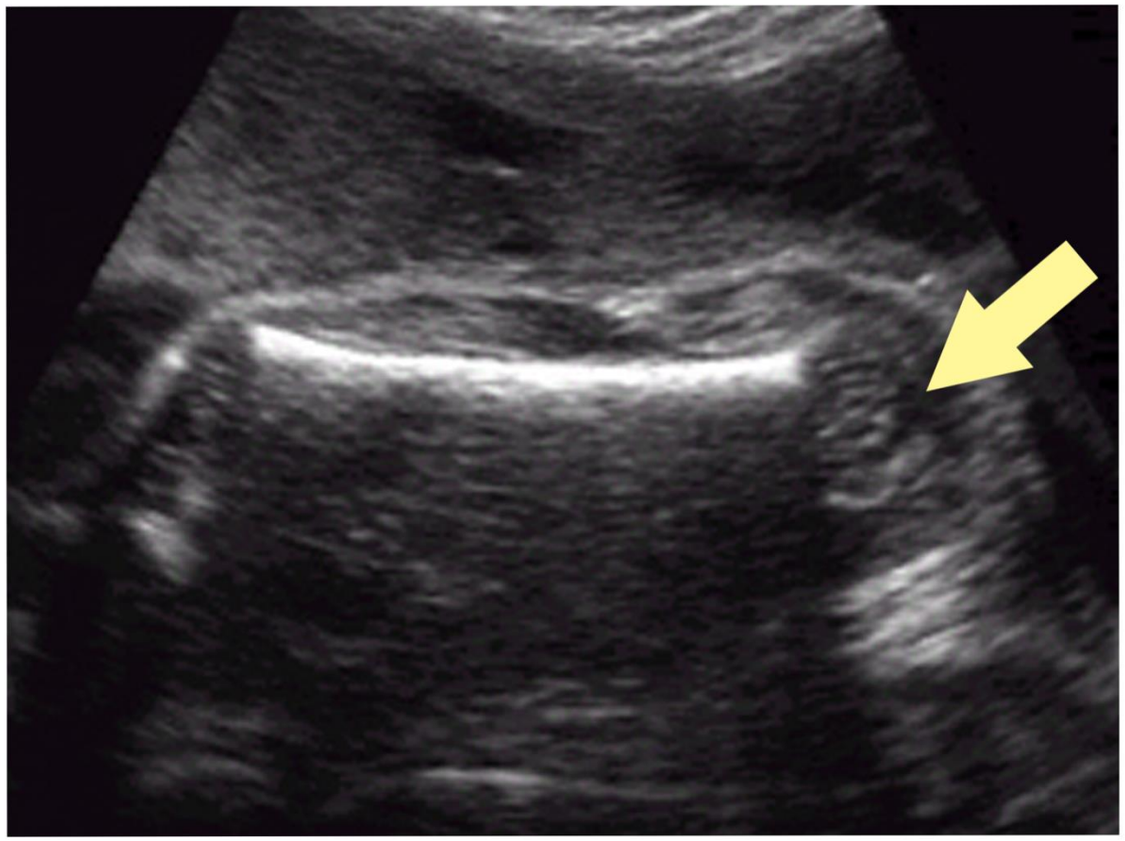
- Lower extremity bones
  - Tibia & fibula & ankle mortise ≈ mid 2nd trimester
  - Metatarsals ≈ 16 weeks
  - Radius & ulna ≈ mid 2nd trimester
  - Femur ≈ mid 2nd trimester

APPENDICULAR SKELETON



Femur

arrow = femoral head

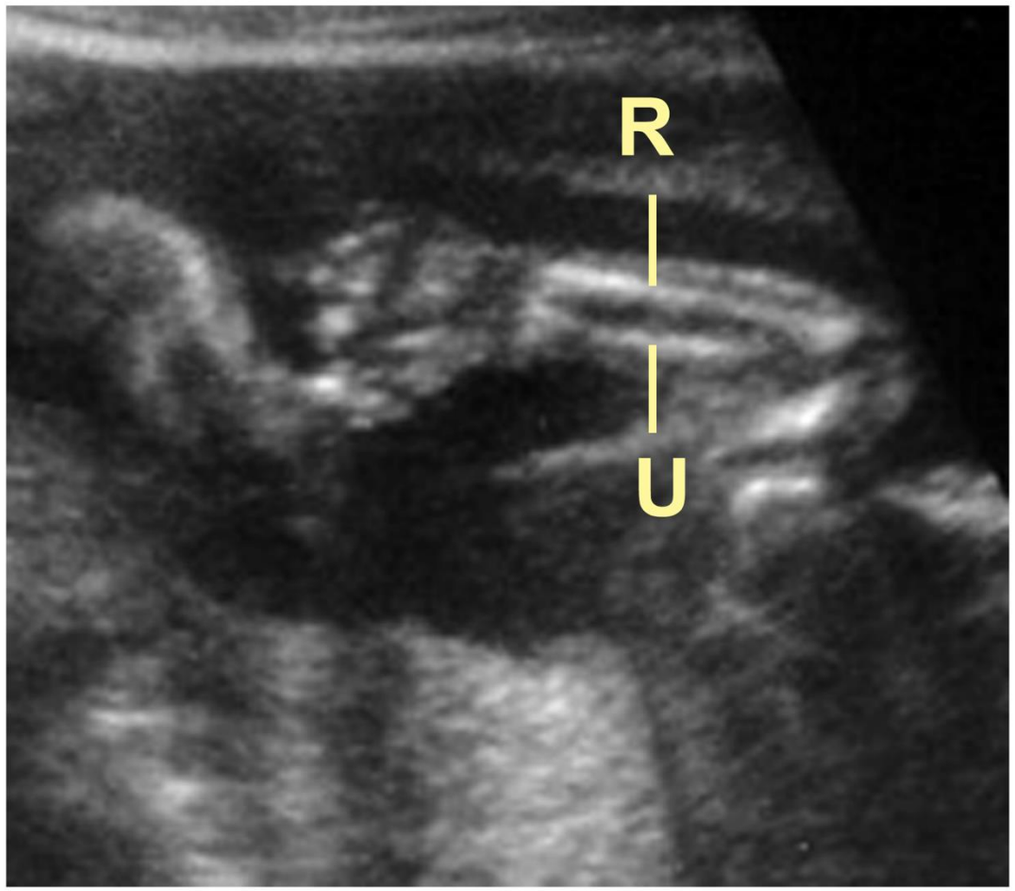


Humerus

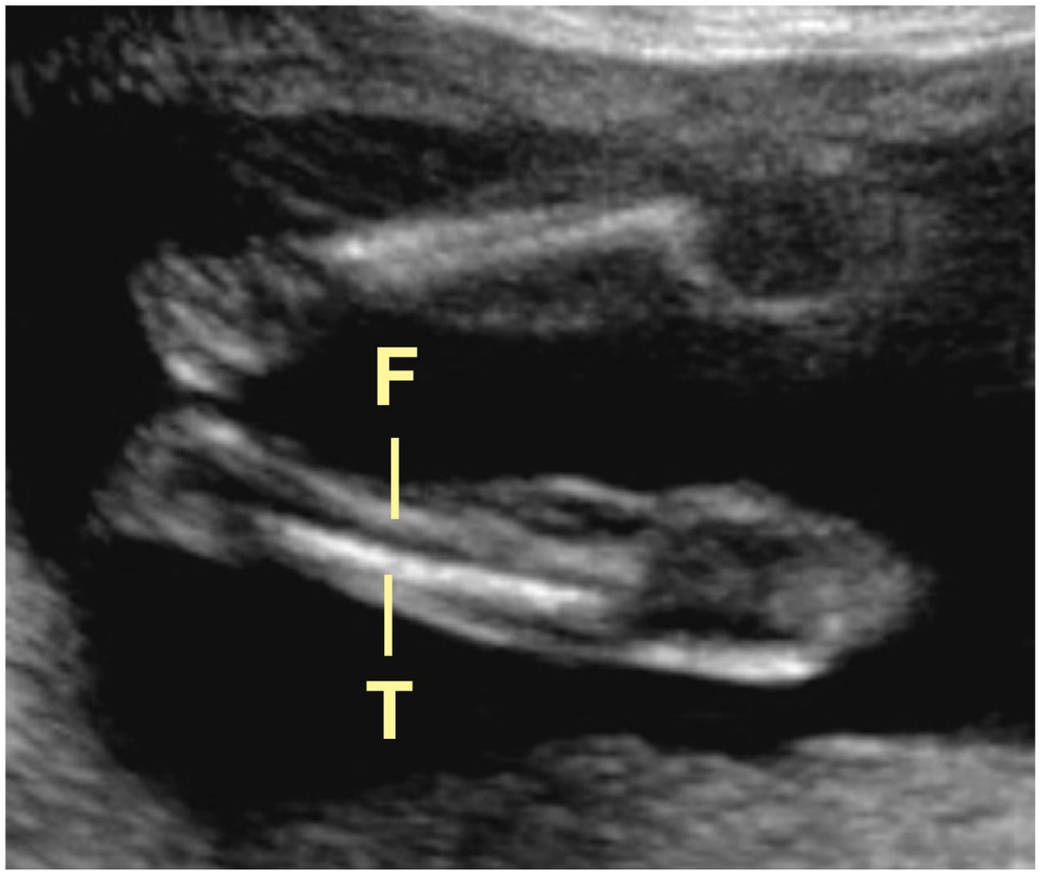
arrow = humeral head

Long bones - proximal

APPENDICULAR SKELETON



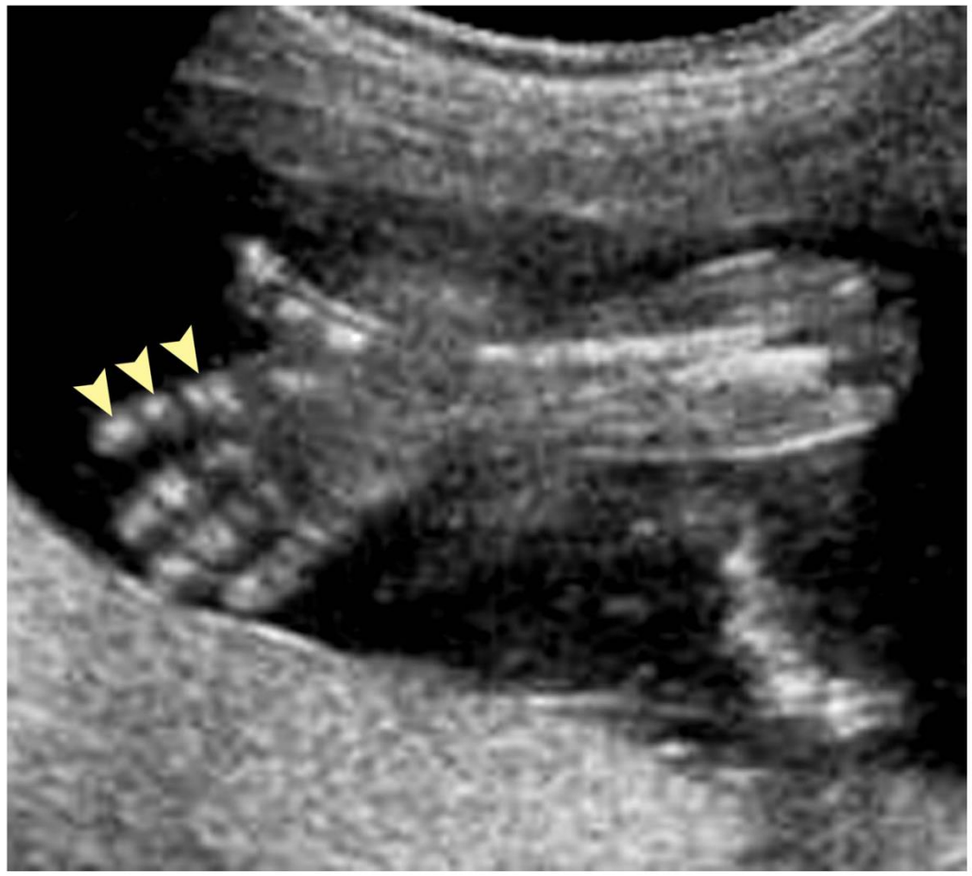
Radius/ulna



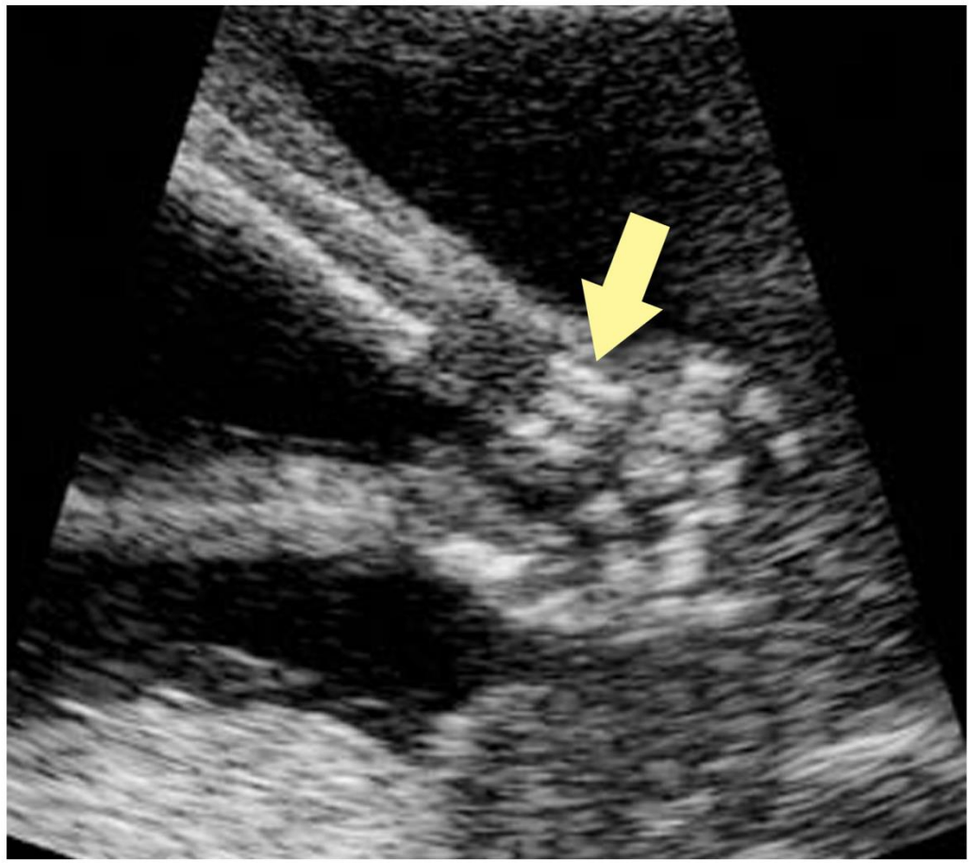
Tibia/fibula

Long bones - distal

APPENDICULAR SKELETON



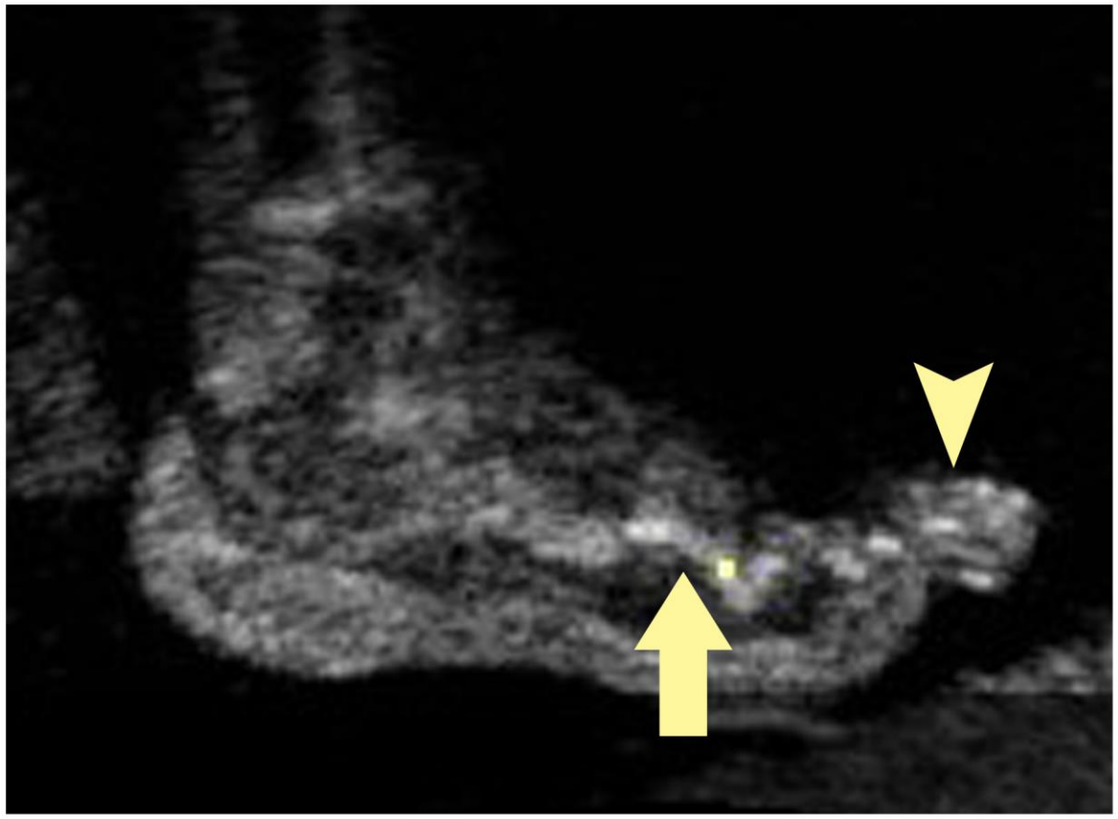
Proximal, mid, distal phalanges



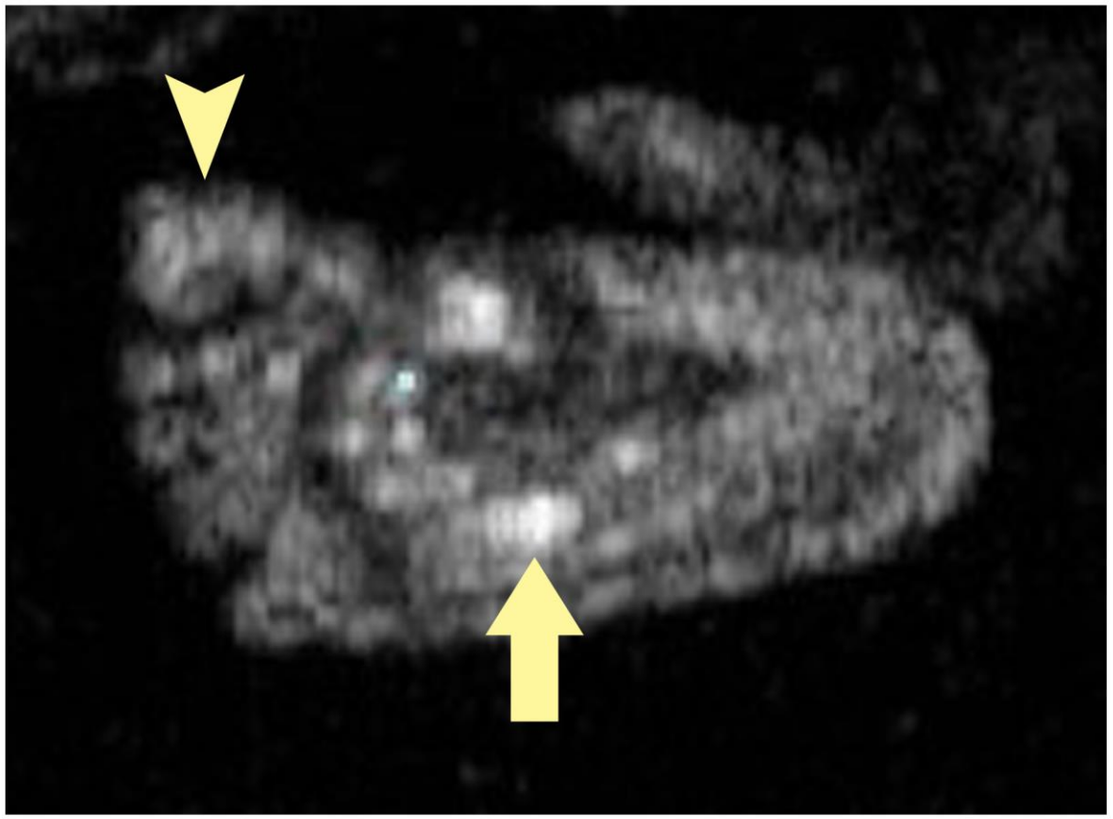
Ossified metacarpals

Upper extremity - hand

APPENDICULAR SKELETON



Foot - lateral view



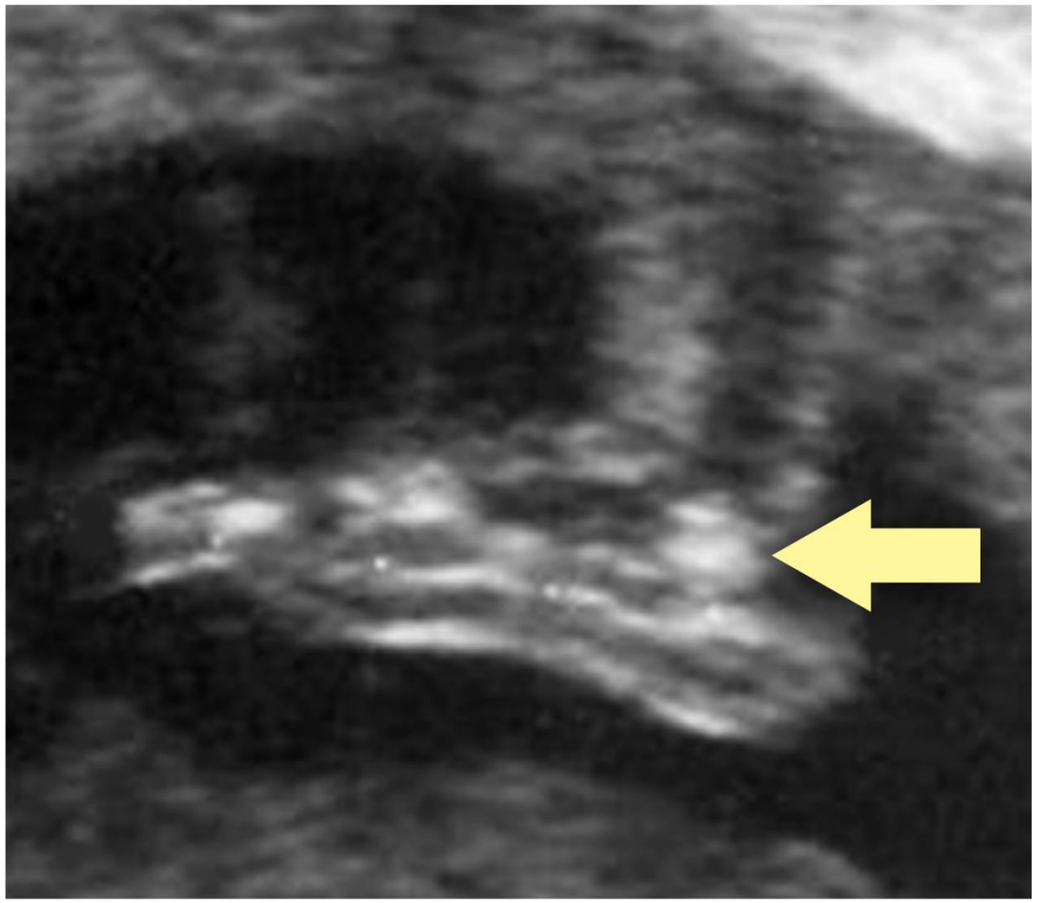
Foot - plantar view

Arrows - metatarsal

Arrowhead = toes

Lower extremity - foot

APPENDICULAR SKELETON



Foot - lateral view

Arrow - calcaneus

Lower extremity - foot

Skeletal Abnormalities

Major Categories

- Osteochondrodysplasias
- Dysostoses
- Idiopathic osteolyses
- Miscellaneous disorders with osseous involvement
- Chromosomal aberrations
- Primary metabolic abnormalities

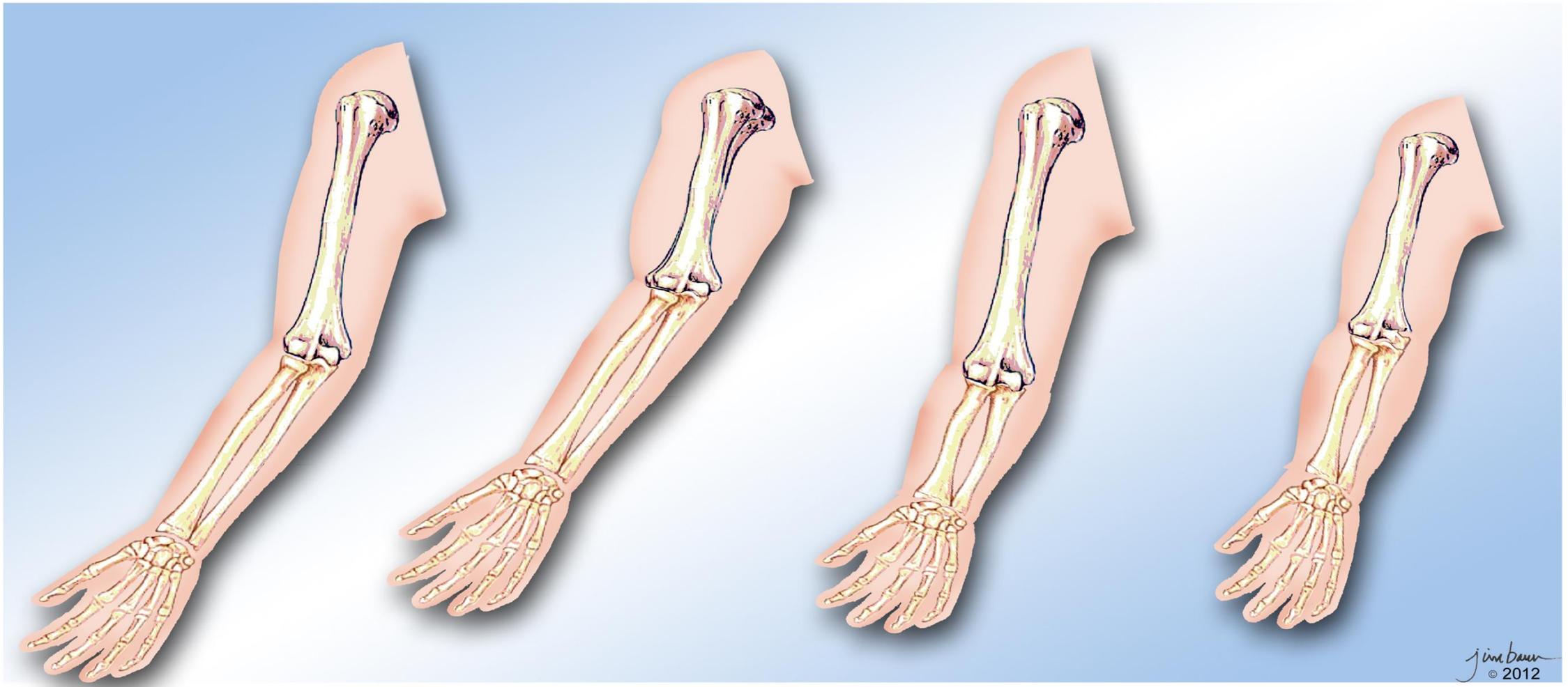
Major Categories

- **Osteochondrodysplasias**
- **Dysostoses**
- **Idiopathic osteolyses**
- Miscellaneous disorders with osseous involvement
- Chromosomal aberrations
- Primary metabolic abnormalities

Definition of Terms

- Most lethal skeletal dysplasias are associated with shortened limbs
- Medical terminology describing type and extend of limb shortening is integral part of preanal sonographic examination
  - *Rhizomelia*: shortening of proximal segment of limb (humerus, femur)
  - *Mesomelia*: Shortening of distal segment of limb (forearm, calf)
  - *Micromelia*: shortening of both proximal and distal segments
  - *Amelia*: absence of a limb

LIMB SHORTENING TERMINOLOGY



Normal **Rhizomelic** **Mesomelic** **Severe Micromelic**

Definition of Terms

- Medical terminology describing type and extend of limb shortening is integral part of preanal sonographic examination
  - *Meromelia*: partial absence of a limb
  - *Polydactyly*: presence of more than five digits on a single hand or foot (fingers or toes)
  - *Syndactyly:* soft tissue or bony fusion of digits (fingers or toes)

LIMB SHORTENING TERMINOLOGY

Meromelia **Polydactyly** **Syndactyly**

Osteochondrodysplasias

- Characterized by:
  - Defects of growth of tubular bones
  - Disorganized development of cartilage and fibrous skeleton
- Sometimes referred to as *dwarf syndromes*
- Severe, lethal type can usually be detected sonographically

Osteochondrodysplasias

- Severe, lethal type can usually be detected sonographically
  - Achondrogenesis
  - Achondroplasia
  - Thanatophoric dysplasia
  - Short rib-polydactyly syndrome
  - Campomelic dysplasia
  - Others

Achondrogenesis

- Rare, lethal form of short-limbed dysplasia
- Virtually no ossification of the vertebral bodies
- May be genetically inherited
- Two types:
  - Type I (Parenti-Fraccaro)
  - Type II (Langer-Saldino)

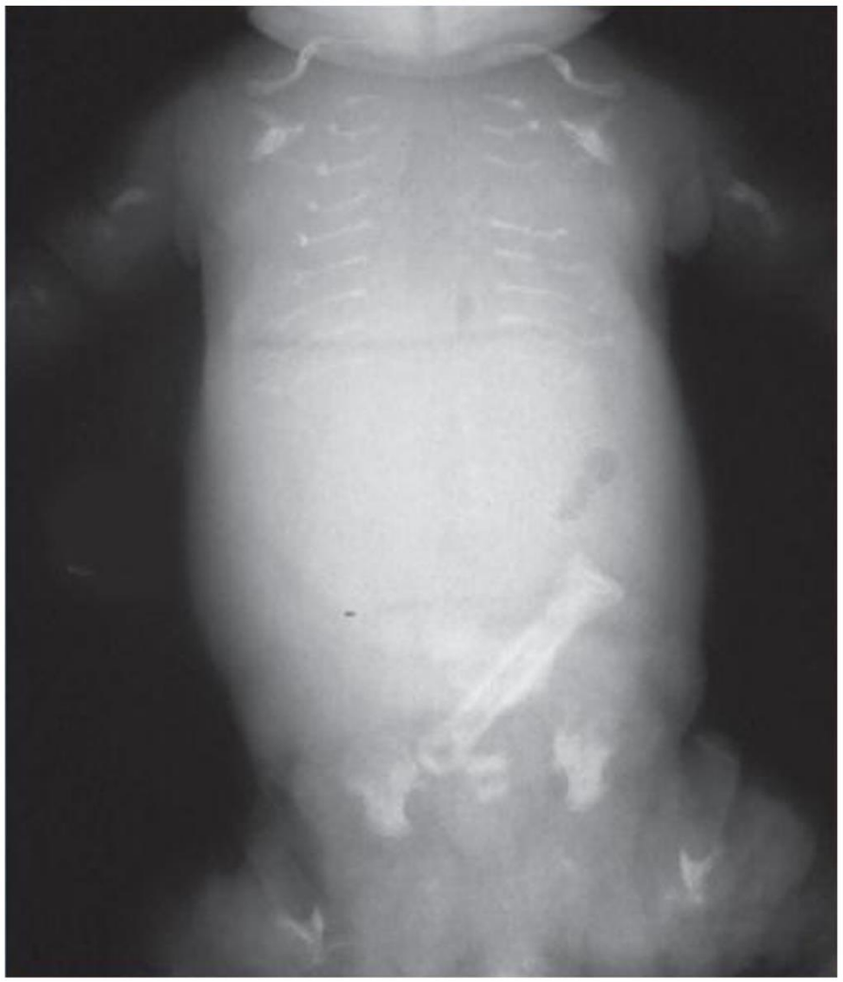
Achondrogenesis

- Associated abnormalities include:
  - IUGR
  - Cleft soft palate
  - Cystic hygroma

Achondrogenesis

- Sonographic findings include:
  - Lack of vertebral ossification ☑
  - Small chest
  - Large head with slightly decreased ossification of cranium (*caput membranaceum*)
  - Severely shortened limbs (*micromelia*)

ACHONDROGENESIS



Postnatal radiograph.

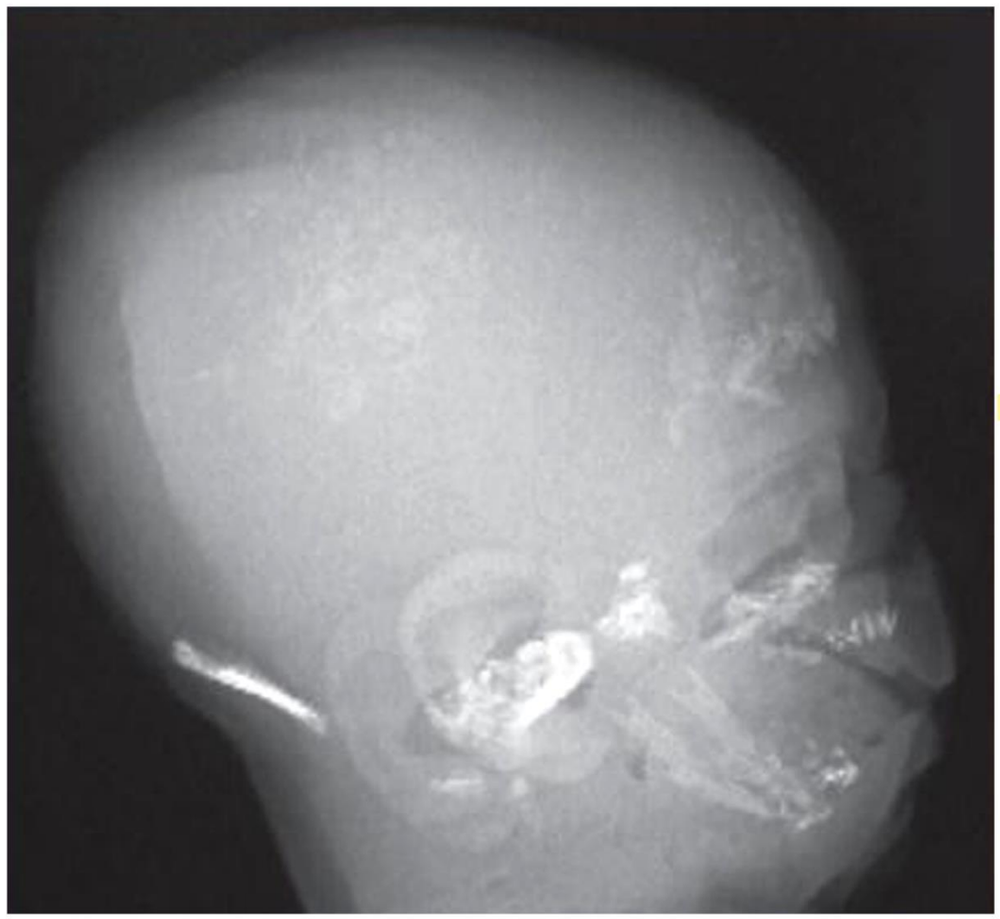
Prenatal sonogram.

Lack of vertebral ossification

ACHONDROGENESIS

Small chest

ACHONDROGENESIS



Postnatal radiograph

Prenatal sonogram

Caput membranaceum

ACHONDROGENESIS

Lower extremity

Upper extremity

Severely shortened limbs

Achondroplasia

- Genetic disorder affecting normal growth and development of skeletal system
- Most common form of short-limbed dysplasia
- Occurs in ≈ 1:25,00 – 50,000 births
- Two types:
  - Heterozygous achondroplasia
  - Homozygous achondroplasia

Heterozygous Achondroplasia

- Nonlethal type
- Characterized by rhizomelic limb shortening after 20 weeks
- Difficult to diagnose prenatally unless one parent has achondroplasia
- Sonographic characteristics:
  - Normal femur length on 1st US (18 – 20 weeks)
  - FL falls below 99th prediction interval by 27 weeks

Homozygous Achondroplasia

- Lethal type
- Both parents are achondroplastic dwarves
- Trait may be carried as autosomal dominant
- Trait may be a spontaneous genetic mutation

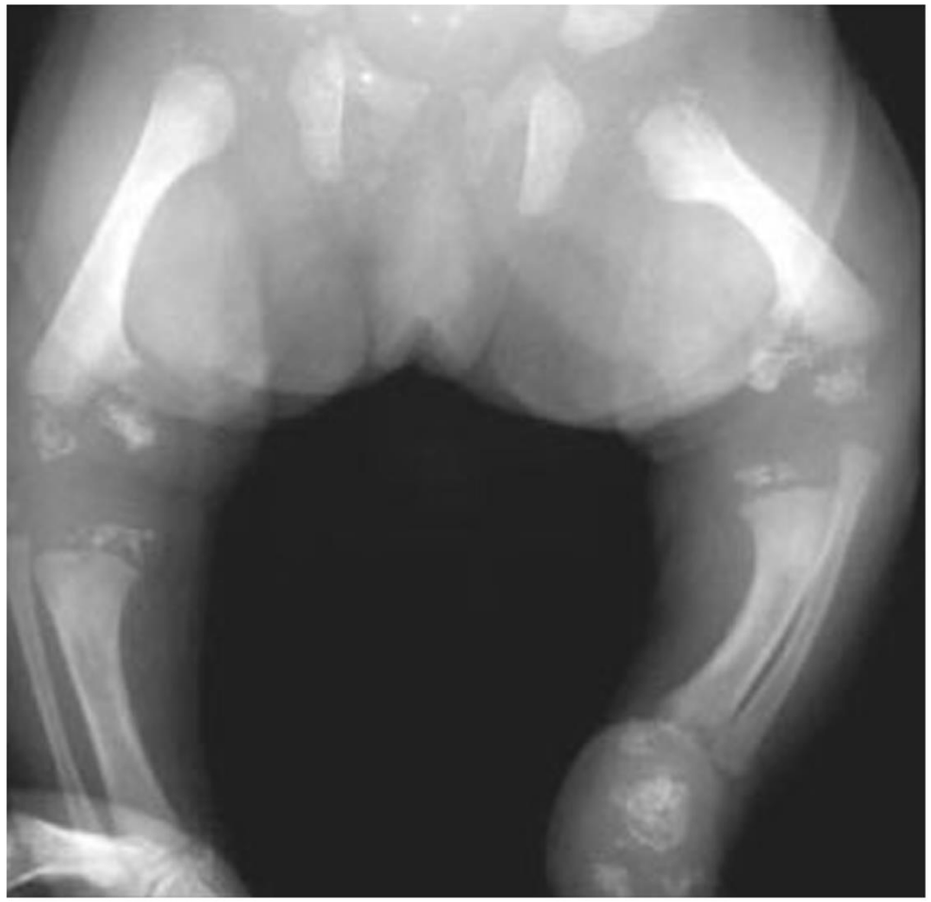
Achondroplasia

- Associated abnormalities with both types include:
  - Macrocephaly
  - Low nasal bridge with prominent forehead
  - Mid-facial hypoplasia
  - Short, tubular bones
  - Trident hand
  - Hydrocephalus
  - Spinal cord compression

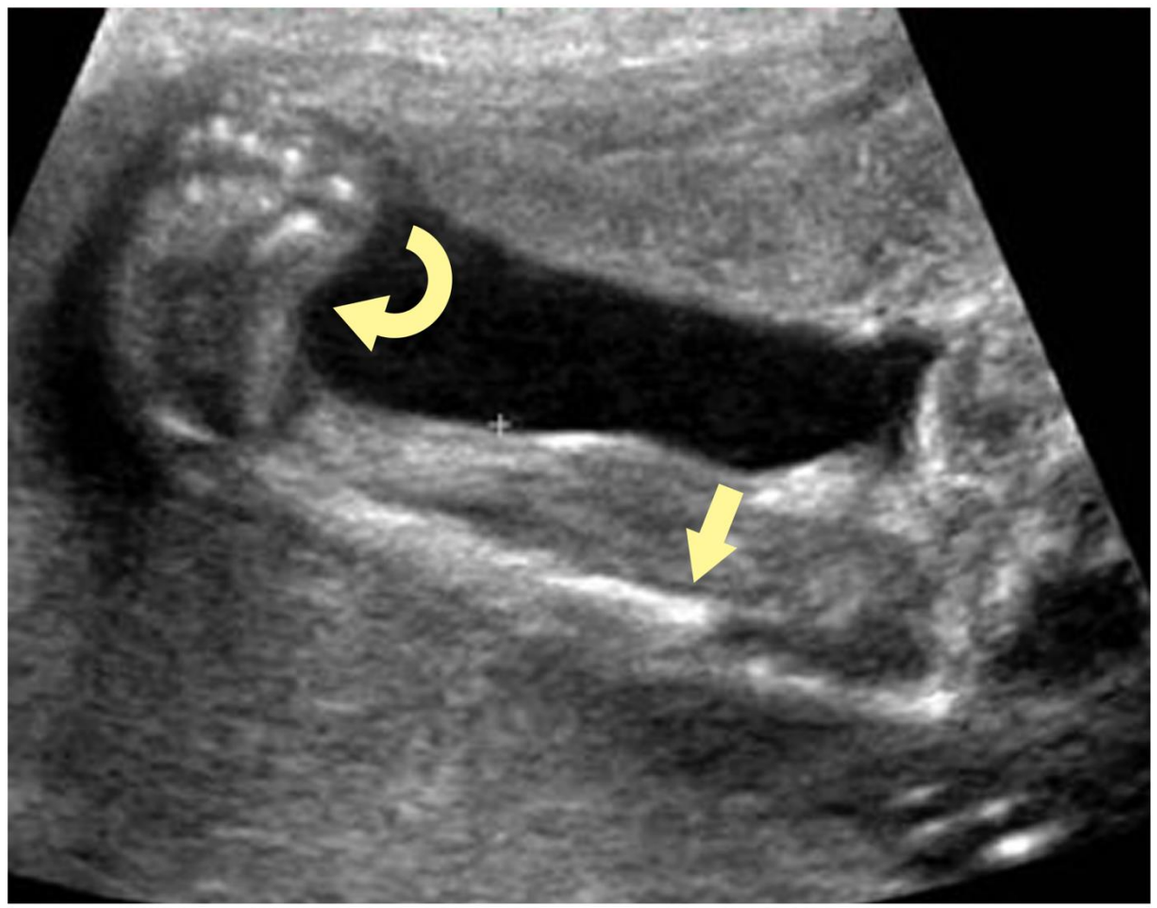
Achondroplasia

- Sonographic findings include:
  - Rhizomelia
  - Frontal bossing of skull
  - Abnormal femur length measurements:
    - Homozygous: FL below 5th percentile prior to 20 weeks
    - Heterozygous: normal FL prior to 20 weeks

ACHONDROPLASIA



Postnatal radiograph.



Prenatal sonogram.

arrow = femur
curved arrow = concomittant talipes equinovarus

Rhizomelia

Thanatophoric Dysplasia

- Lethal skeletal dysplasia characterized by:
  - Cloverleaf skull
  - Both parents of normal stature
  - Extreme rhizomelia
  - Short, bowed limbs
  - Hypoplastic thorax

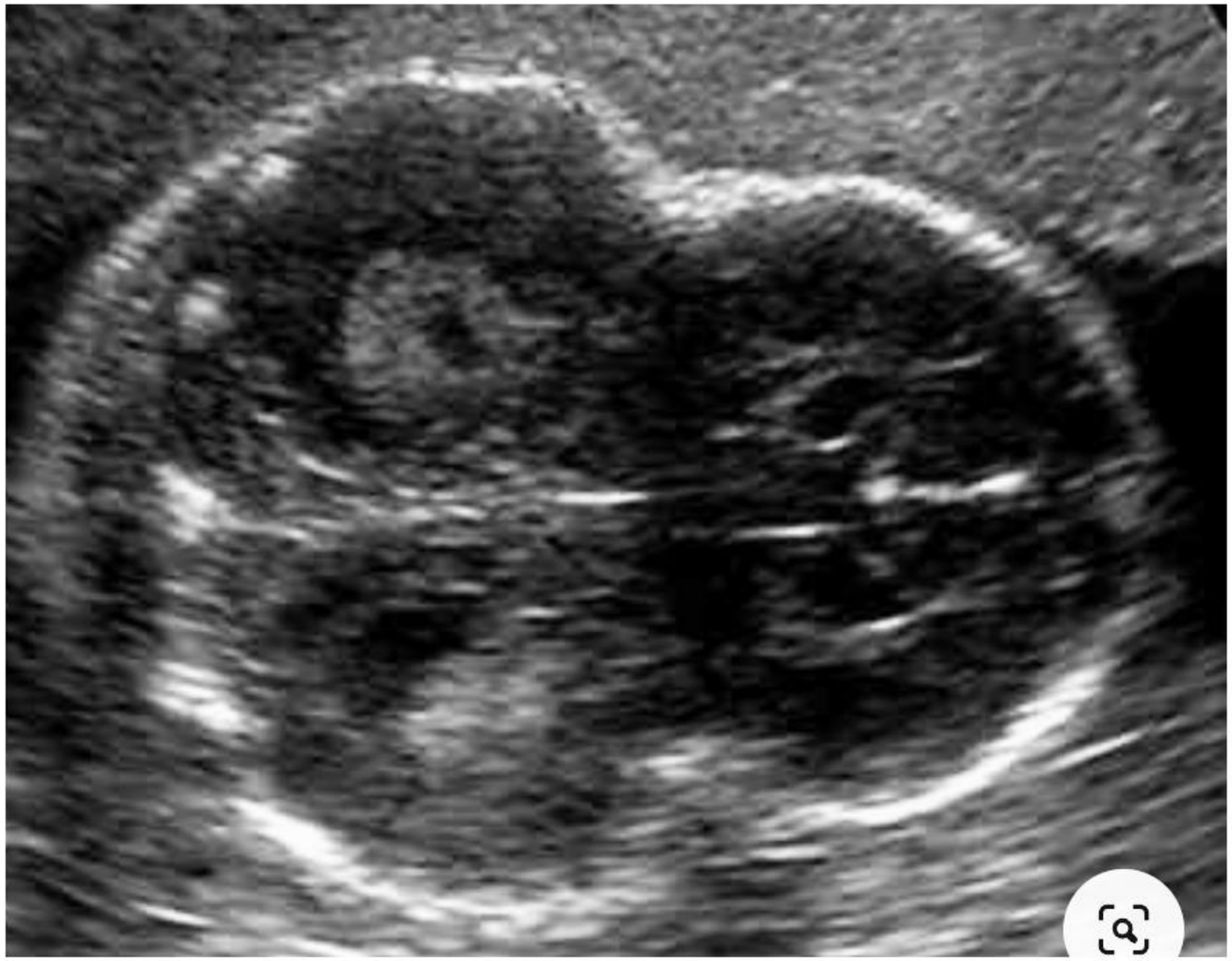
Thanatophoric Dysplasia

- Associated abnormalities include:
  - Macrocephaly
  - Hydrocephalus
  - Patent ductus arteriosus
  - Atrial septal defect
  - Horseshoe kidney
  - Hydronephrosis
  - Imperforate anus

Thanatophoric Dysplasia

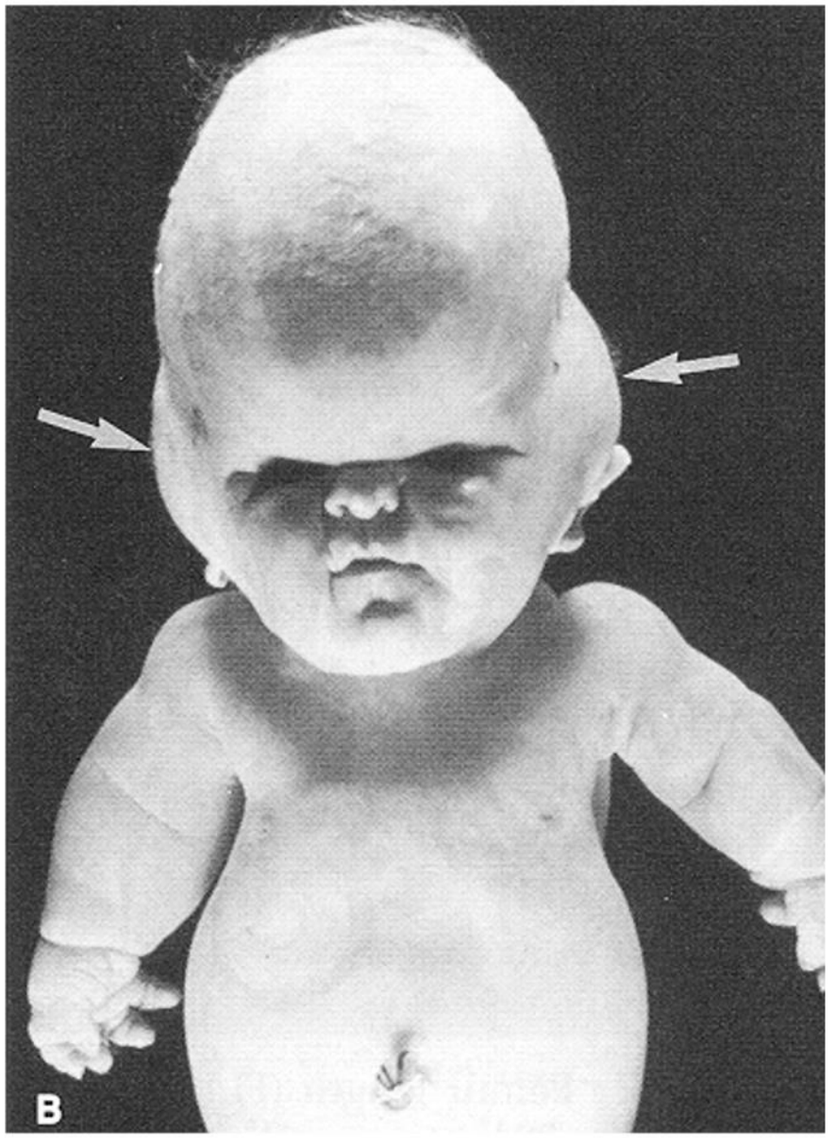
- Sonographic findings include:
  - Cloverleaf skull ☑
  - Markedly short and densely ossified, bowed, long bones
  - Hypoplastic thorax (bell-shaped chest)
  - Trident hand deformity
  - Polyhydramnios (71% of cases)
  - Flattened vertebral bodies

THANATOPHORIC DYSPLASIA



Cloverleaf skull

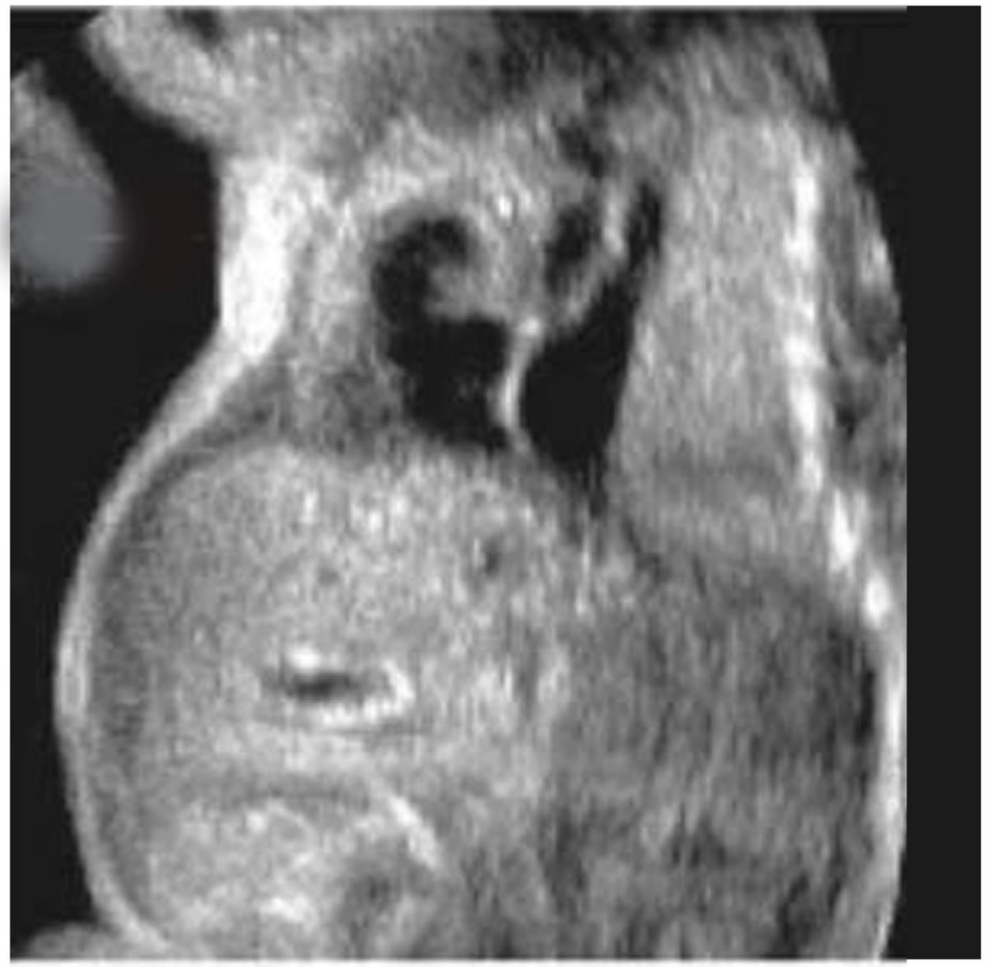
THANATOPHORIC DYSPLASIA



Cloverleaf skull

THANATOPHORIC DYSPLASIA

Sagittal section



Coronal section

Hypoplastic thorax

THANATOPHORIC DYSPLASIA

Trident hand deformity

Short Rib-Polydactyly Syndrome

- Lethal skeletal dysplasia characterized by:
  - Short limbs (*micromelia*)
  - Excessive number of digits (*polydactyly*)
  - Extremely narrowed thorax

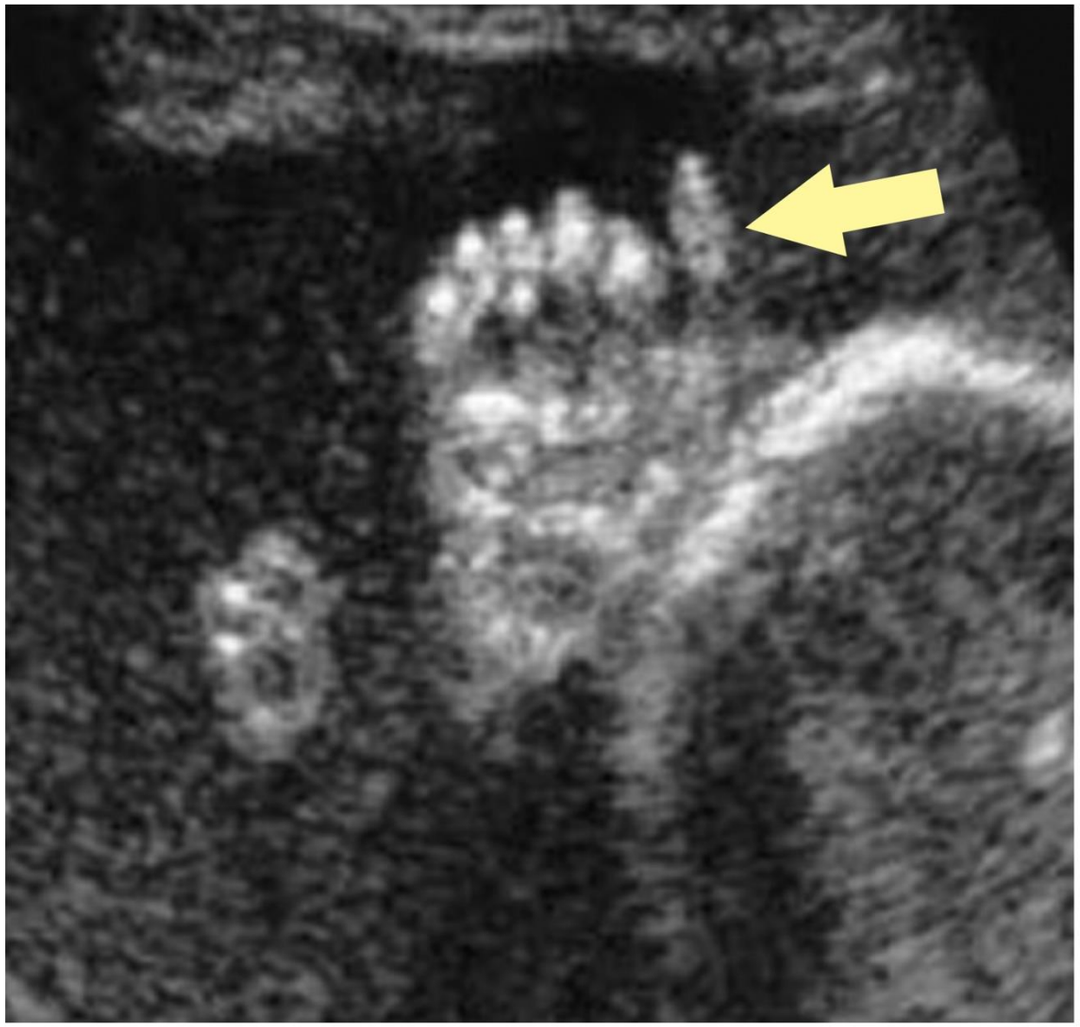
Short Rib-Polydactyly Syndrome

- Associated abnormalities include:
  - Cardiac defects
  - Polycystic kidneys
  - Imperforate anus

Short Rib-Polydactyly Syndrome

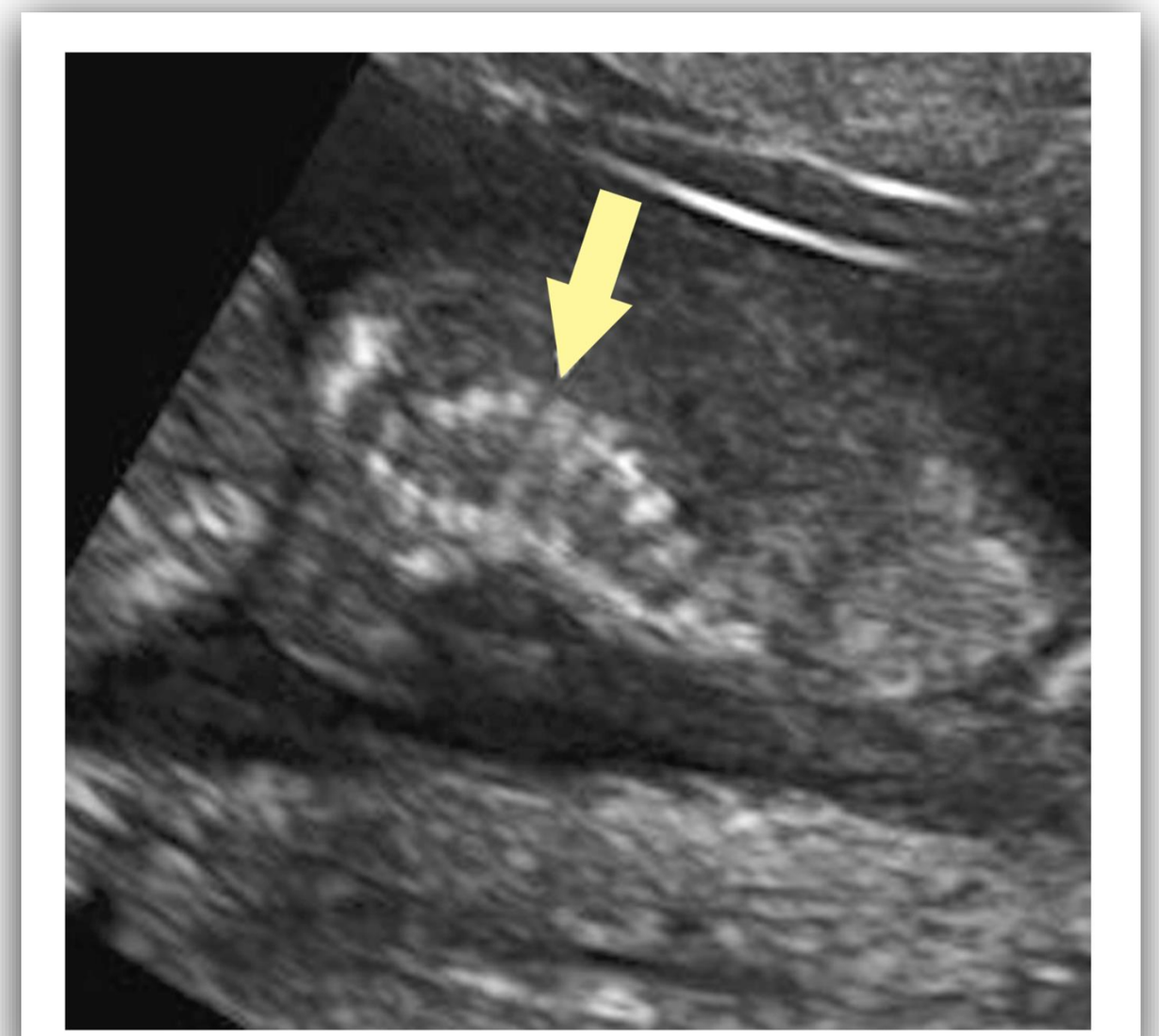
- Sonographic findings include:
  - Polydactyly ☑
  - Narrowed thorax
  - Striking micromelia
  - Choroid plexus cysts

SHORT RIB-POLYDACTYLY SYNDROME



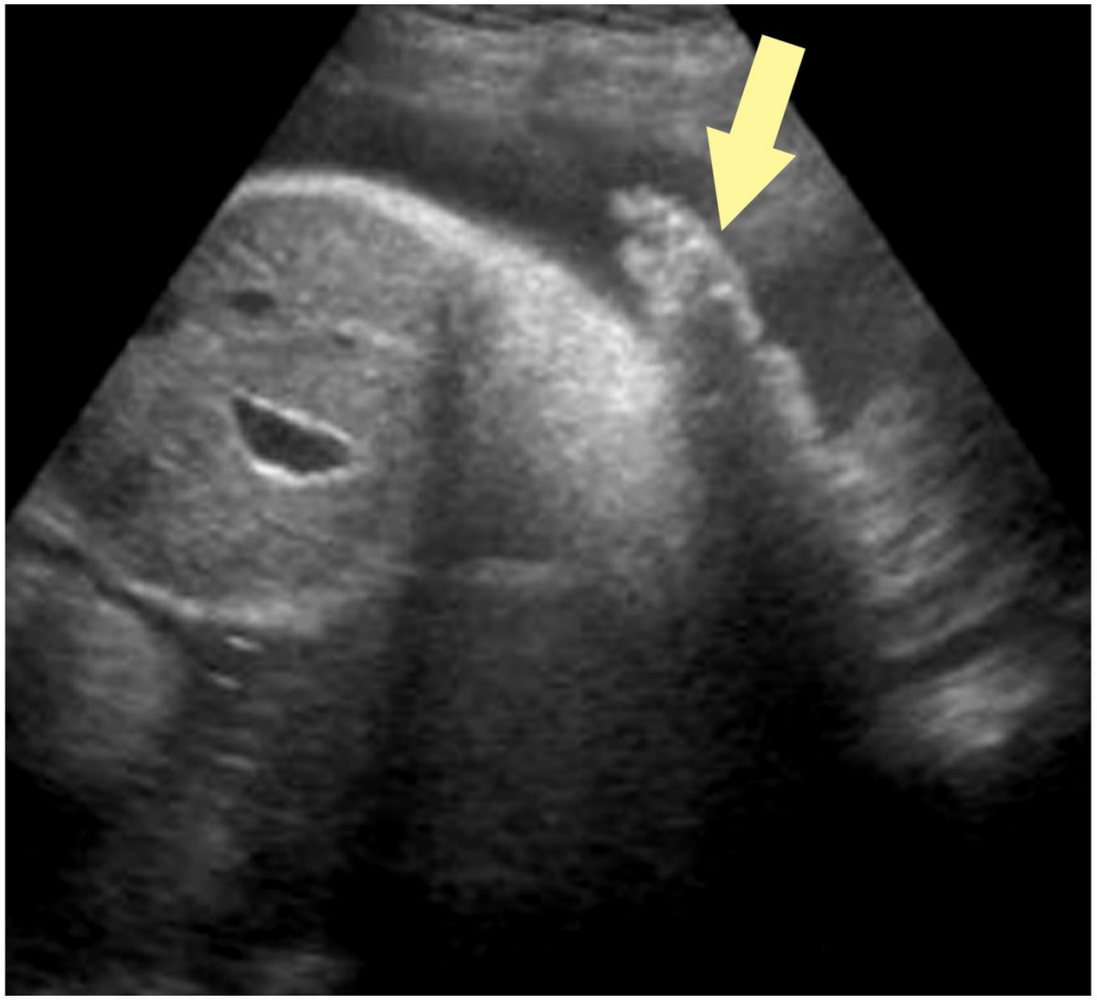
Polydactyly

SHORT RIB-POLYDACTYLY SYNDROME



Narrowed thorax

SHORT RIB-POLYDACTYLY SYNDROME



Striking micromelia

Campomelic Dysplasia

- Not always lethal
- *Campomelia* from French *bent limbs*
- Characterized by:
  - Bowing of long bones esp. lower extremity
  - Club feet are common
  - Hydronephrosis
  - Hydrocephaly

Campomelic Dysplasia

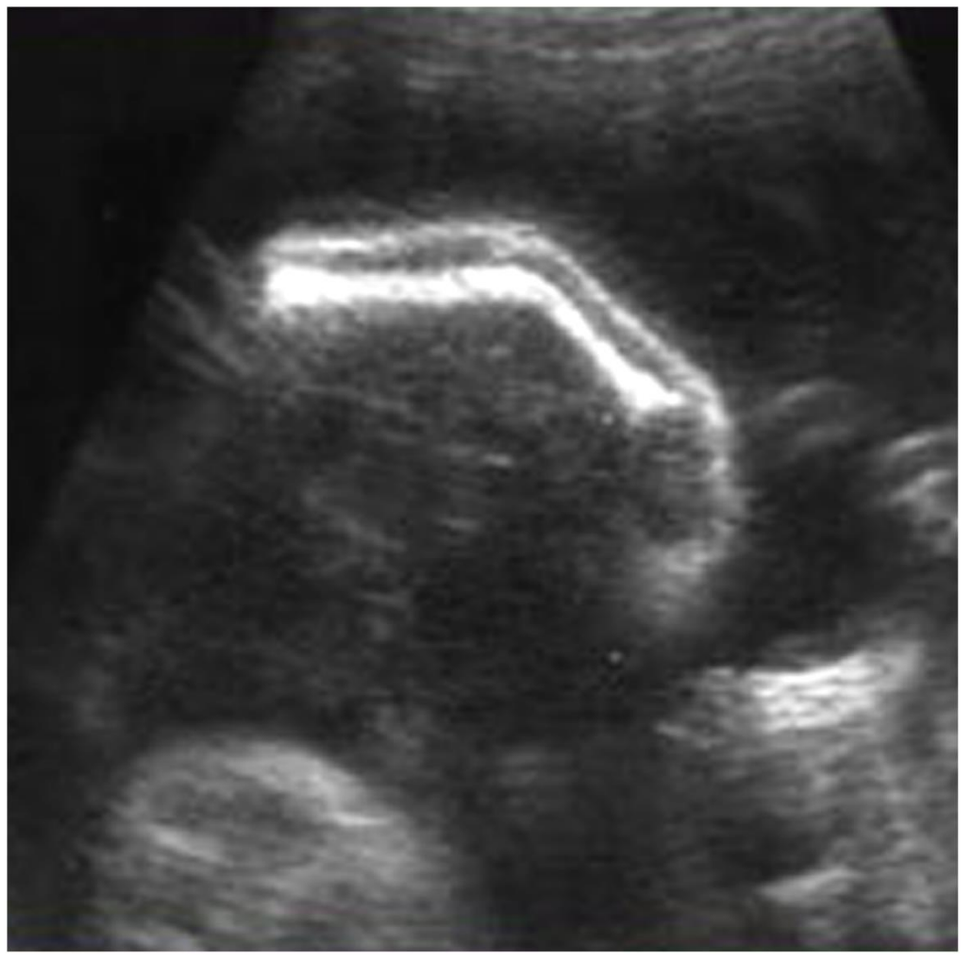
- Associated abnormalities include:
  - Hydrocephalus
  - GU dysgenesis
  - Micrognathia
  - Cardiovascular abnormalities
  - Polyhydramnios

Campomelic Dysplasia

- Sonographic findings include:
  - Sever bowing of long bones
  - Especially in lower extremities
  - Narrowed thorax
  - Associated hydronephrosis or hydrocephalus
  - Possible clubfoot

CAMPOMELIC DYSPLASIA

Postnatal radiograph



Prenatal sonogram

Severe bowing of long bones

CAMPOMELIC DYSPLASIA

Narrowed thorax

CAMPOMELIC DYSPLASIA

Bowed tibia and clubfoot

Idiopathic Osteolyses

- Autosomal dominant disorders characterized by failure of normal by ossification process
- Most common types encountered prenatally:
  - Osteogenesis imperfecta (OI)
  - Hypophosphatasia

Osteogenesis Imperfecta

- Disorder of collagen production, secretion, or function
- Eight types ranging from mild to lethal
- Overriding characteristics:
  - Hypomineralization of bone
  - Abnormal fragility of skeletal structures
  - *In utero* fractures result in bone shortening
  - Risk for delivery trauma resulting in intracranial hemorrhage and stillbirth

Osteogenesis Imperfecta

- Associated abnormalities include:
  - IUGR
  - Macrocephaly
  - Umbilical hernia

Hypophosphatasia

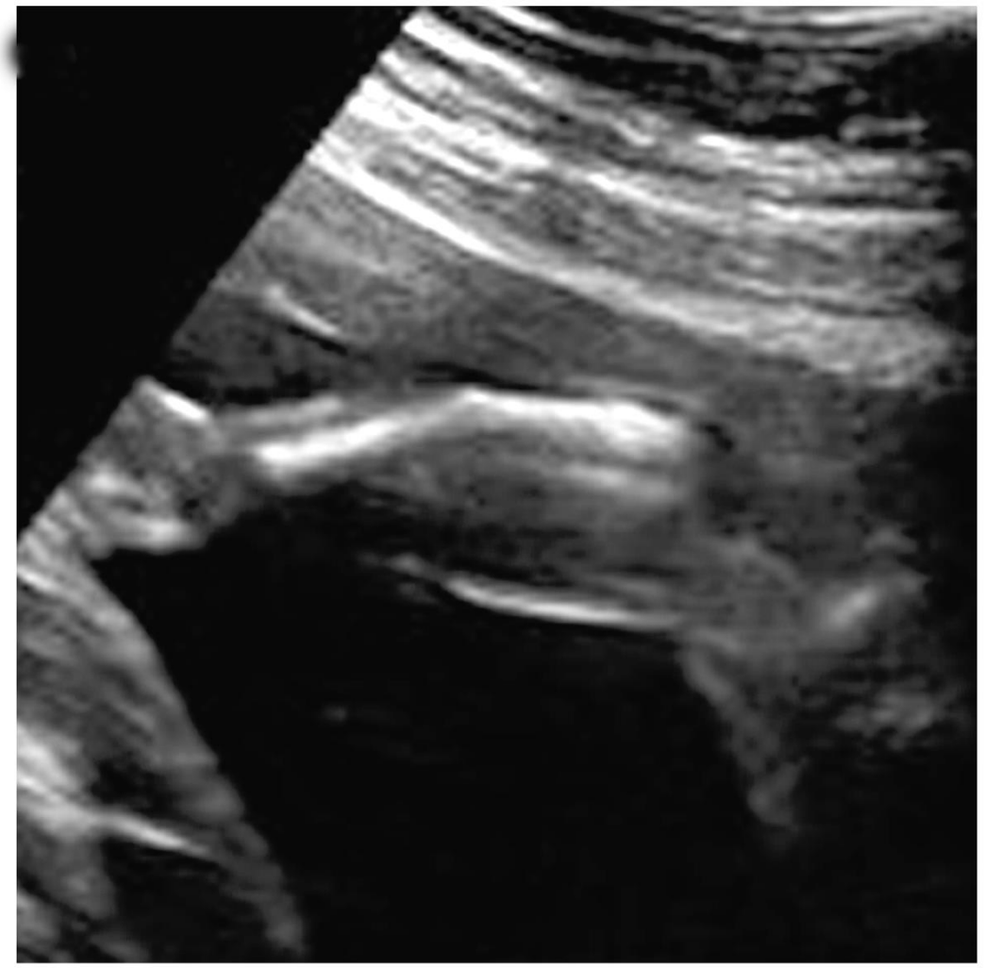
- Similar in manifestation to OI
- Deficiency of serum alkaline phosphatase (not collagen)
- Several subtypes. *Perinatal* subtype is uniformly lethal
- Overriding characteristics are similar to OI:
  - Hypomineralization of bone
  - Abnormal fragility of skeletal structures
  - *In utero* fractures result in bone shortening

Idiopathic Osteolyses

- Sonographic findings include:
  - Presence of long bone fractures or excessive callus formation ☑
  - Drastically shortened long-bones with bowing
  - Hypomineralized skeletal structures
  - Enhanced resolution of intracranial anatomy
  - Decreased calvarial ossification the permit pressure deformity with transducer compression
  - Rib cage deformities

IDIOPATHIC OSTEOLYSES

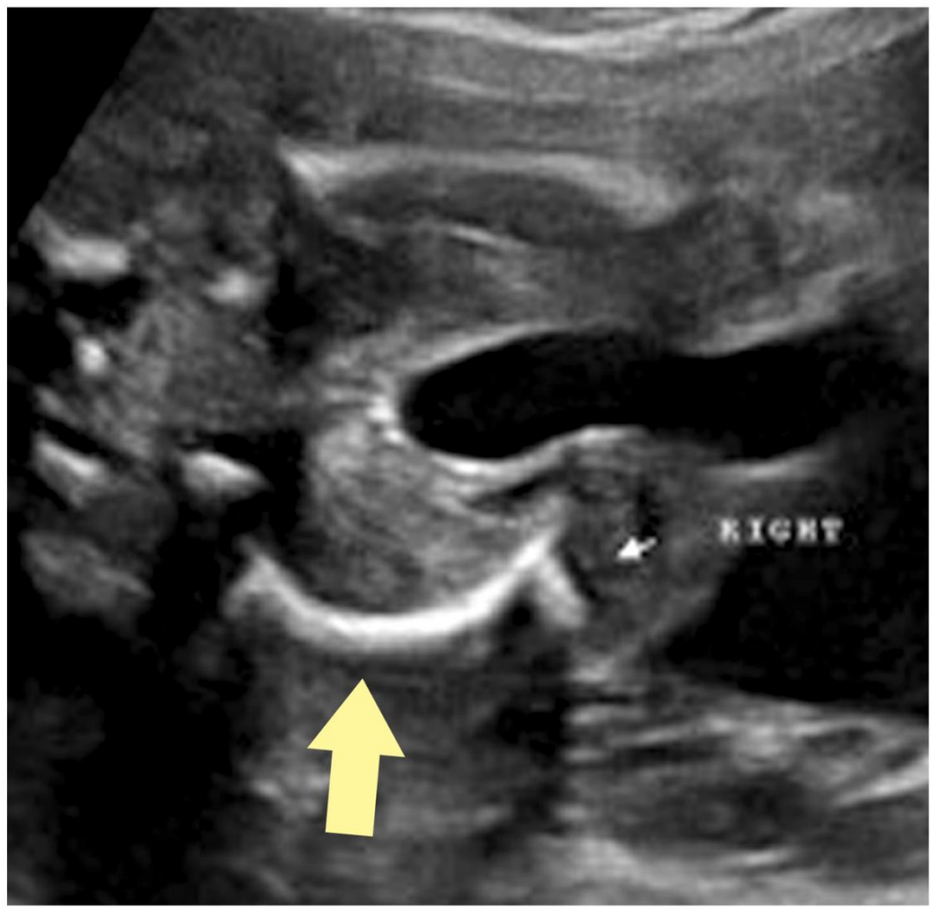
Tibial fracture



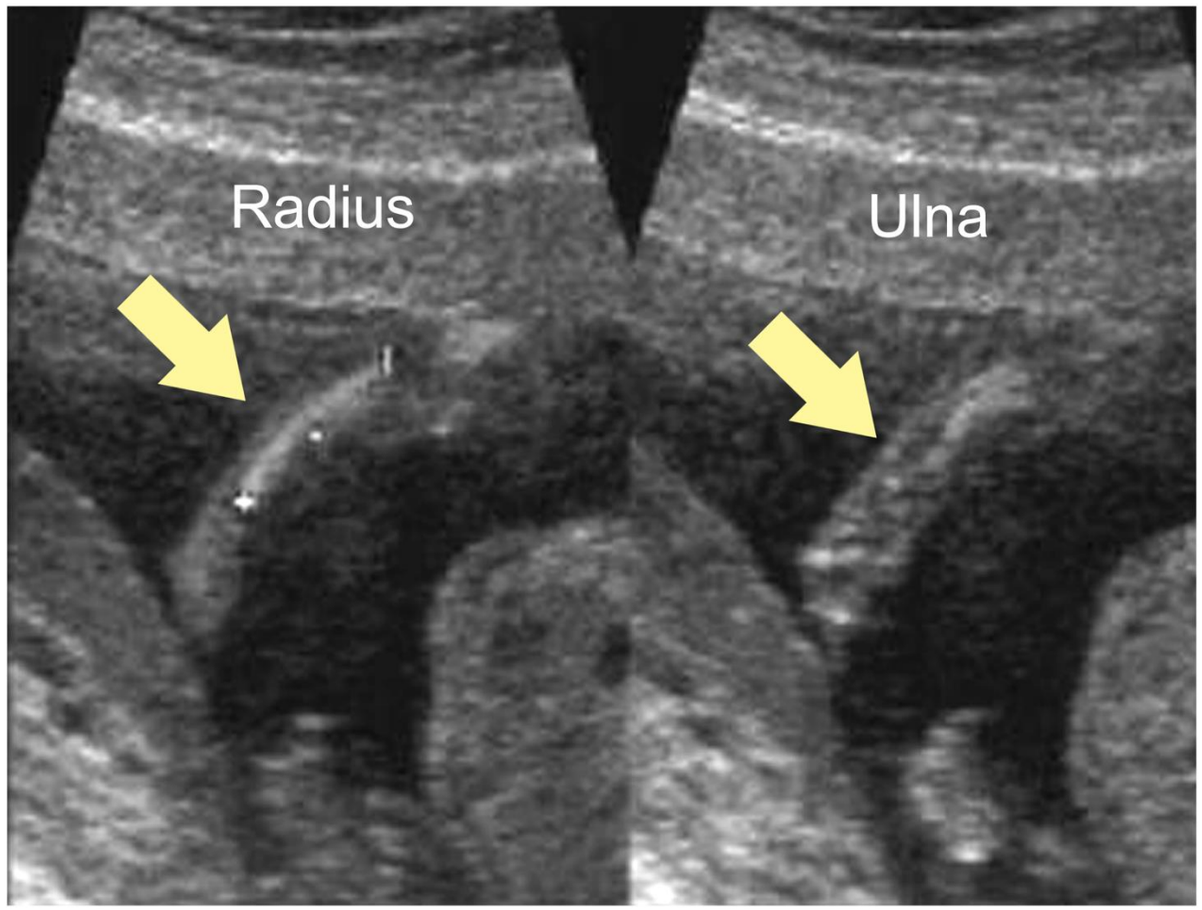
Radial fracture

Long-bone fractures

IDIOPATHIC OSTEOLYSES



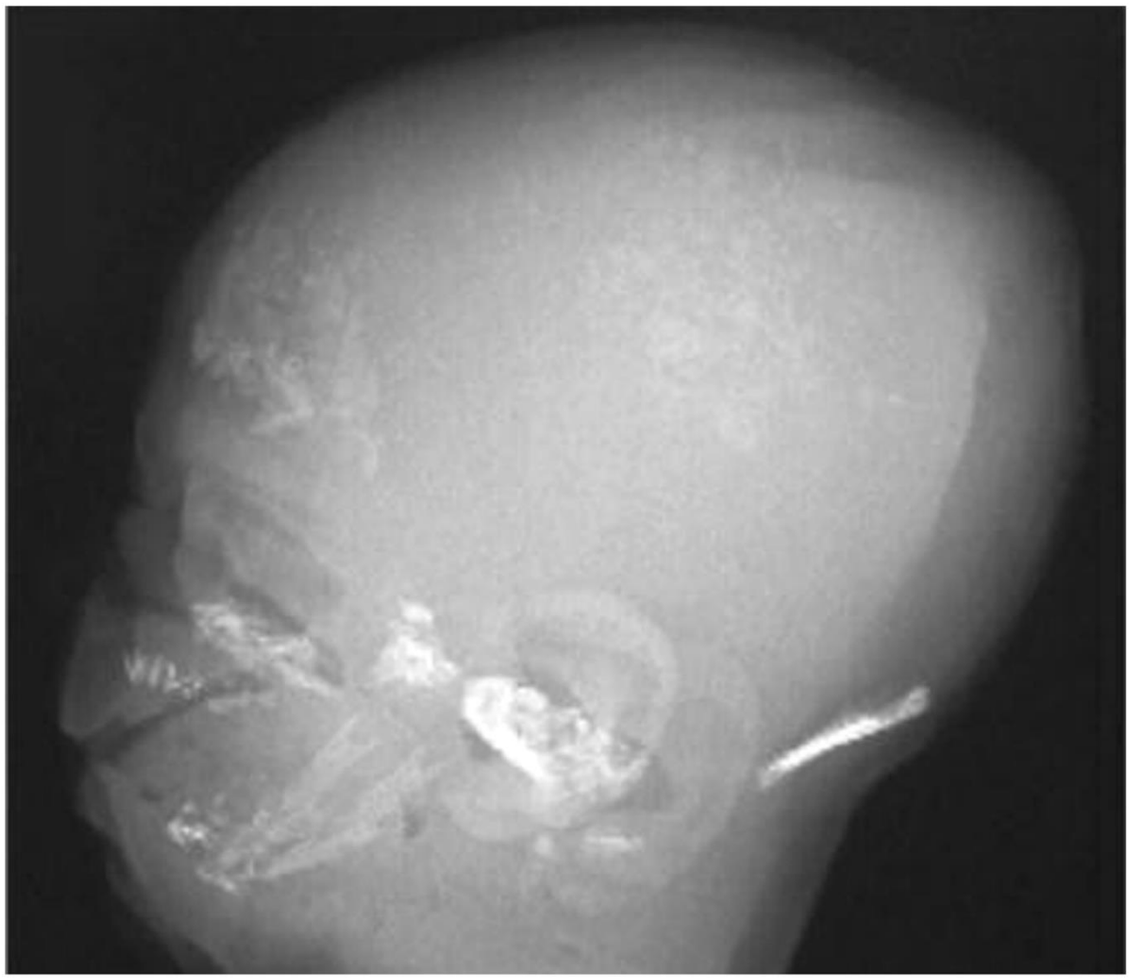
Femur



Radius and ulna

Drastically shortened long-bones with bowing

IDIOPATHIC OSTEOLYSES



Postnatal radiograph

Prenatal sonogram absent acoustic shadowing

Hypomineralized skeletal structures

IDIOPATHIC OSTEOLYSES

Enhanced resolution of intracranial anatomy

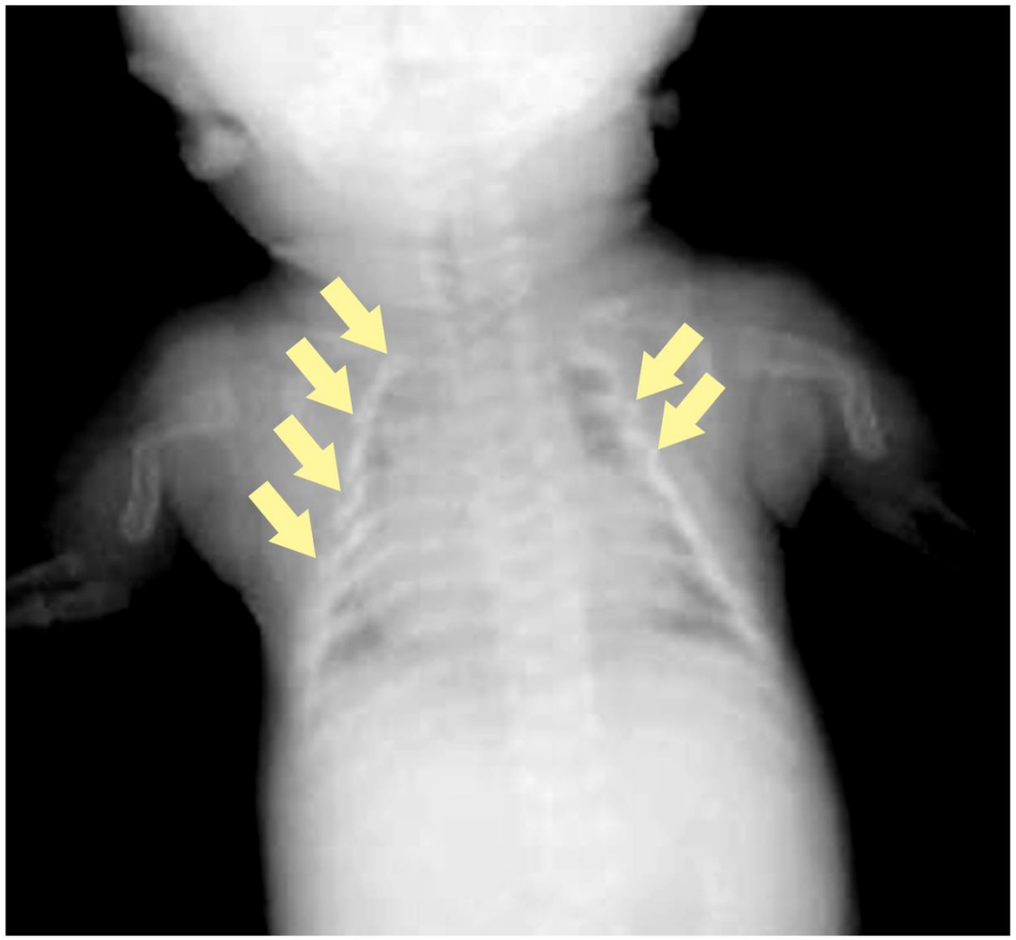
IDIOPATHIC OSTEOLYSES

Focal deformity from transabdominal transducer pressure

Spontaneous cranial deformity by fundal placenta and oligohydramnios

Pressure deformities

IDIOPATHIC OSTEOLYSES



Postnatal radiograph demonstratng multiple bilateral rib fractures

Prenatal sonogram

Rib cage deformities

Dysostoses

- Isolated bony malformation that may occur alone or in conjunction with various syndromes
- Most common types encountered prenatally:
  - Talipes equinovarus (*clubfoot*)
  - Rocker bottom foot
  - Radial ray anomaly

Talipes Equinovarus

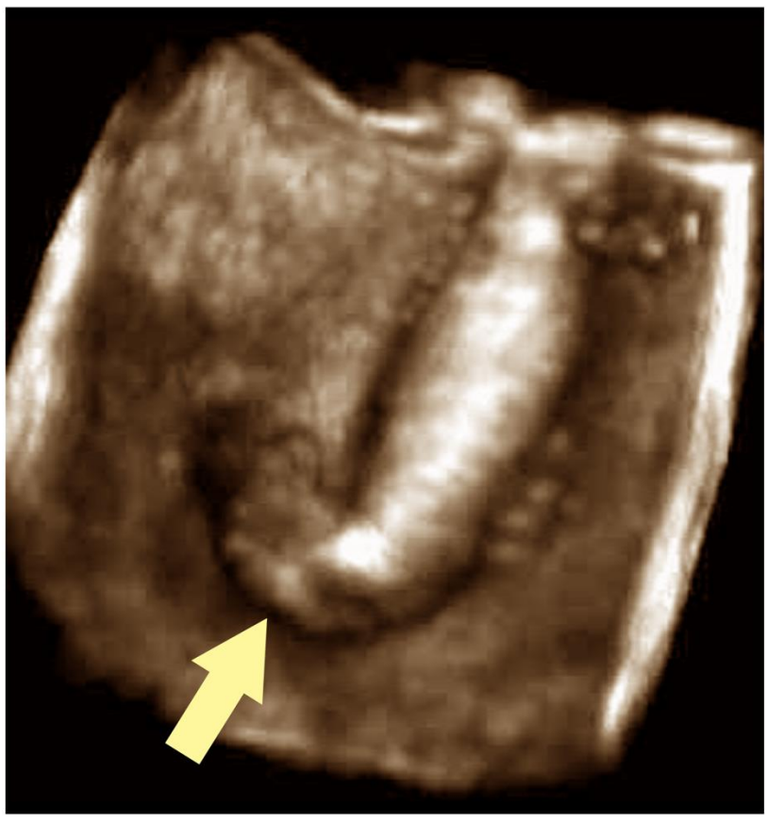
- Most common skeletal anomaly detected during routine OB sonographic examination
- Can occur as an isolated defect or in conjunction with multiple syndromes
- Etiologies include:
  - Genetic
  - Environmental
  - Uterine constraint (*amniotic band syndrome*)

Talipes Equinovarus

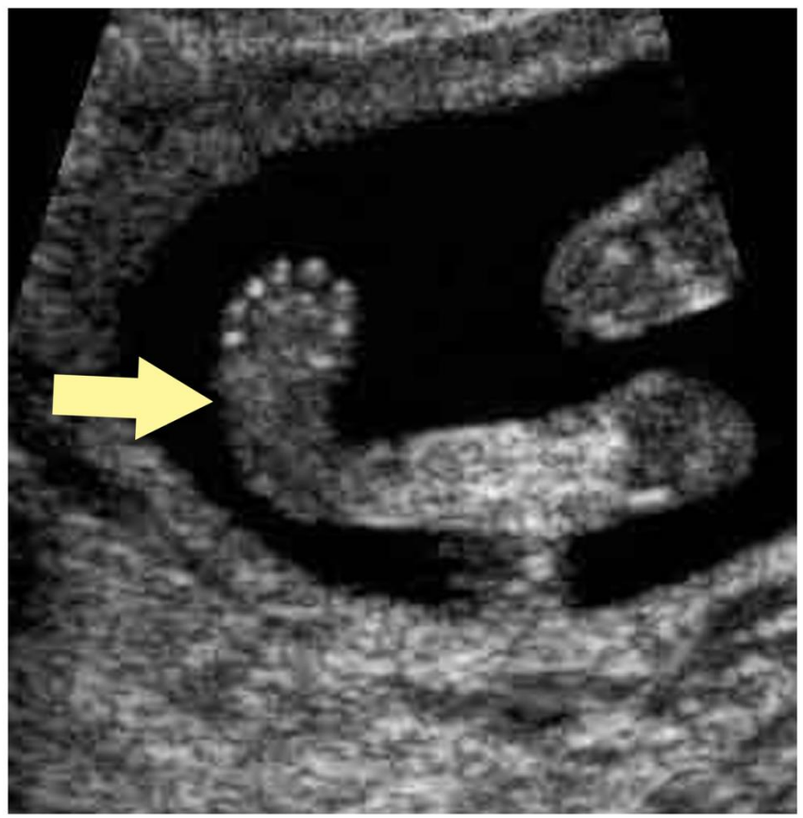
- Score of associated anomalous conditions (*too many to list*)
- Pathology: inversion of foot and flexion of sole
- Sonographic detection based on knowledge of relative orientation of foot and lower extremity
  - Normal: lateral view of tib-fib = lateral view of foot
  - Clubfoot: lateral view of tib-fib = foot appears AP

TALIPES EQUINOVARUS

Normal feet



Clubfoot



Clubfoot (standard 2D imaging)

TALIPES EQUINOVARUS

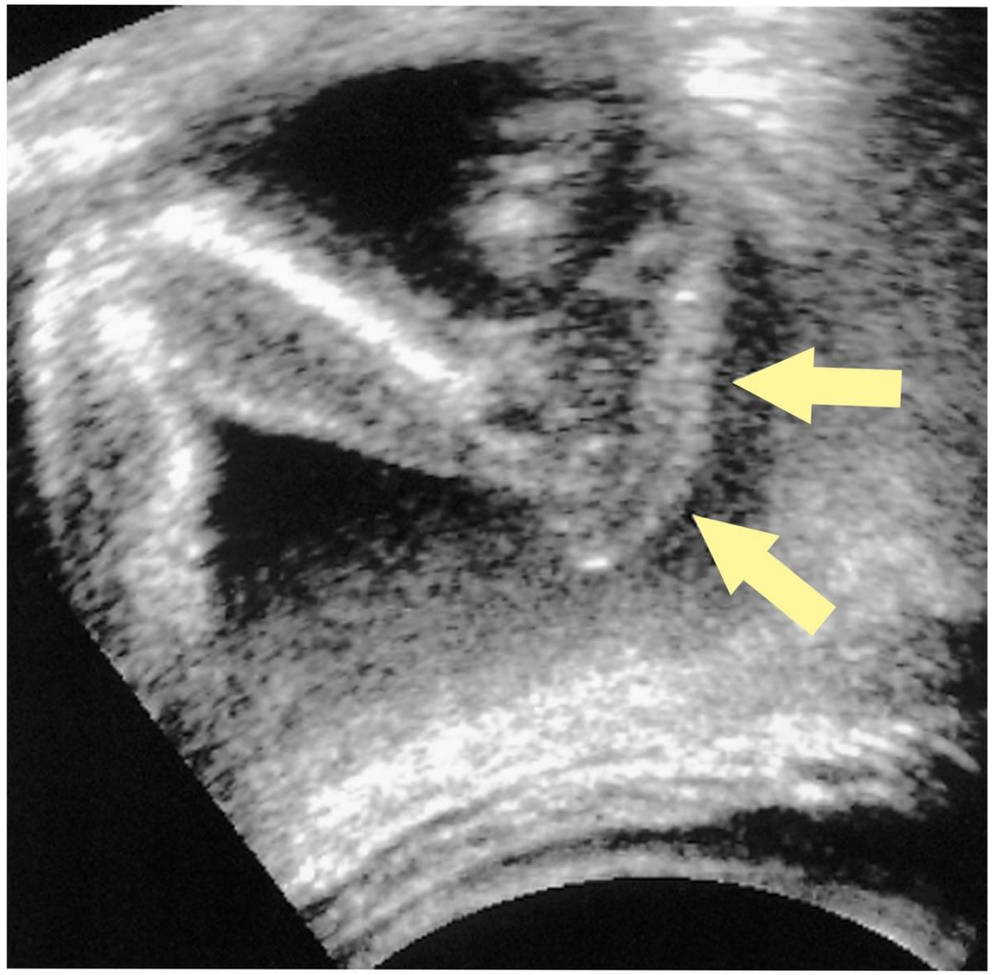
Bilateral

Rocker Bottom Foot

- Dorsal and lateral dislocation of the talonavicular joint and prominent calcaneus
- Mimics appearance of the rocker on a rocking chair
- Sometimes classified as soft sign for aneuploidy
  - Trisomy 13 (Patau syndrome)
  - Trisomy 18 (Edwards syndrome)
  - 18q deletion syndrome
  - Spina bifida

ROCKER BOTTOM FOOT

Radiograph



Sonographic demonstration

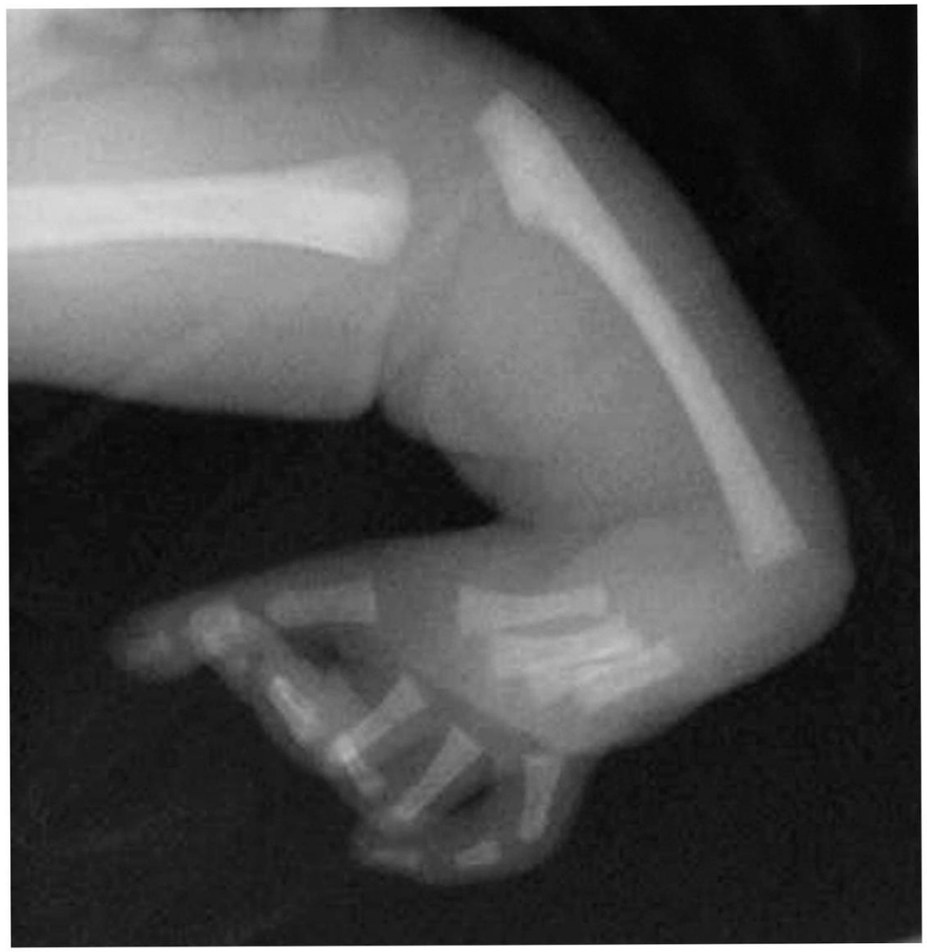
Radial Ray Anomaly

- Partial to complete absence of the radius
- Usually associated with abnormalities of wrist and thumb
- Large spectrum of appearances
- Associated abnormalities include:
  - Trisomy 18 (Edward syndrome)
  - Amniotic band syndrome
  - Holt-Oram syndrome
  - VACTERL association

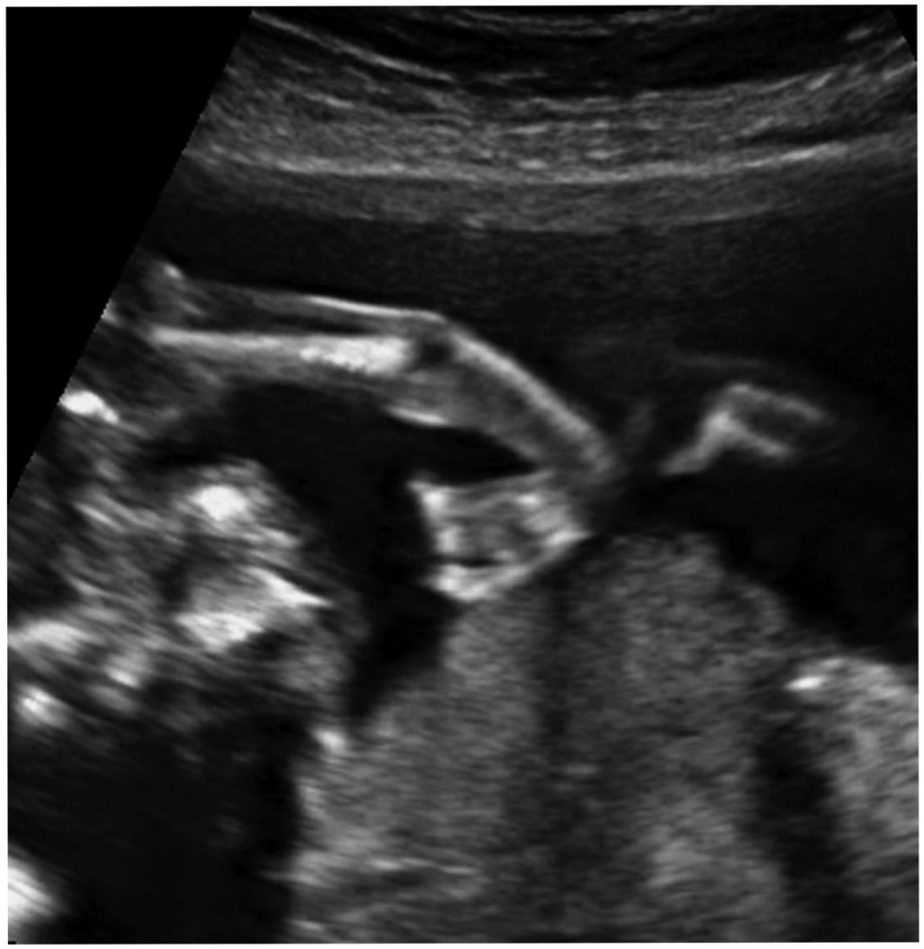
Radial Ray Anomaly

- Sonographic findings include:
  - Absent or hypoplastic radius
  - Sharp medial rotation of hand
  - Absent thumb in some cases

RADIAL RAY ANOMALY



Postnatal radiograph



Prenatal sonogram

OB GYN SONOGRAPHY REVIEW

Fetal Skeleton