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Fetal Anatomy: Skeletal

Normal Anatomy

1. General Considerations
   a. By 15-16 weeks most bones can be imaged. Ossification center is visualized, not entire structure which contains cartilaginous tissue.

2. Appendicular Skeleton
   a. Imaged well by early-mid second trimester.
   b. Long bones are easily seen including metacarpals, metatarsals and phalanges.
   c. Carpals are not ossified until after birth, therefore they are not seen. Exception is calcaneus which ossifies between 5-6 month.
   d. Scapula and clavicle can be seen.

3. Axial Skeleton:
   a. Definition:
   b. Skull: cranium, facial bones, pelvis, spine. Sphenoid bone and petrous ridges seen at base of skull separating cranial fossae.
   c. Facial bones: orbits, maxilla, mandible and bony nasal septum.
   d. Pelvis: iliac ossification centers seen from early second trimester. Ischial ossification centers seen at about 20 weeks.
   e. Spine: can be seen with great clarity especially after 22 weeks. Transverse image offer best method of evaluation.
      i. Composed of three ossification centers; two posterior and one anterior. On longitudinal the posterior elements are seen as parallel bands.

Skeletal Anomalies

1. **Classification of Skeletal Anomalies:** **T**here are six major categories of skeletal anomalies:
   a. Osteochondrodysplasias
      i. Defects of growth of tubular bones
      ii. Disorganized development of cartilage and fibrous skeleton
      iii. Abnormalities of density of cortex

b. Dysostoses
    i. With cranial and facial involvement
    ii. With predominant axial involvement
    iii. With predominant involvement of extremities
c. Idiopathic osteolyses
    i. Osteogenesis imperfecta
    ii. Hypophosphatasia
d. Miscellaneous disorders with osseous involvement
e. Chromosomal aberrations
f. Primary metabolic abnormalities

2. Osteochondrodysplasias

a. **Pathology**: Also referred to as dwarf syndromes. Abnormalities of cartilage and/or bone growth and development. Characterized by defects of tubular bone growth. Of the vast number of variations in this group of anomalies, only a few are identifiable pre-natally with ultrasound. Fortunately, the severe conditions that are lethal can usually be detected sonographically. The most common lethal skeletal dysplasias are described below.

b. **Achondrogenesis**
    i. Definition: A rare, lethal form of short-limbed dysplasia which may be inherited genetically. Pathologically it is a failure of ossification process. Two types:
    ii. TYPE I: (Parenti-Fraccaro)
        1. Absent vertebral ossification centers
        2. Incomplete ossification of skull
        3. Rib fractures
        4. Arms extremely short and stubby
        5. Head not enlarged compared to trunk
    iii. TYPE II: (Langer-Saldino)
        1. Head large compared to body
        2. Prominent skin folds over a short neck
        3. Small chest
        4. Distended abdomen and possible fetal hydrops
        5. Very short limbs held away from body
    iv. Sonographic findings:
        1. Lack of vertebral ossification
        2. Large head with slightly decreased ossification of the cranium.
        3. Severely shortened-limbs (usually involves all limbs)
        4. Small chest

c. **Homozygous Dominant Achondroplasia**
    i. Definition: A lethal, short limbed dysplasia characterized by rhizomelic dwarfism, limb bowing, lordotic spine and a bulky head. It occurs in fetuses in which both parents are achondroplastic dwarfs.

ii. Sonographic Findings:
1. Both parents are achondroplastic dwarfs
2. Cloverleaf skull
3. Shortened long bones in 3rd trimester
4. Femur/BPD ratio below 1st percentile

d. **Heterozygous Achondroplasia**
i. Definition: A non-lethal dysplasia characterized by rhizomelic shortening of the limbs and drop off of femur length after 20 weeks. In 80% of cases, a spontaneous genetic mutation is the cause. In some cases, the trait is carried as autosomal dominant.
ii. Sonographic Findings:
1. By 27 weeks, femur lengths fall below 99 percent prediction interval
2. Rhizomelia
3. Normal femur length prior to 20 weeks

e. **Thanatophoric Dysplasia**
i. Definition: Lethal skeletal dysplasia characterized by extreme rhizomelia, bowed long bones, narrow thorax with normal trunk length and a relatively large head. Severely flattened vertebral bodies. Thorax is narrow and respiratory distress usually follows birth leading to death. Pathologically, this condition is associated with numerous anomalies including cloverleaf skull, horseshoe kidney, atrial septal defects (ASD), imperforate anus. Cloverleaf skull results from premature closure of coronal and lambdoidal sutures and is pathognomonic for this condition.
ii. Sonographic Findings:
1. Cloverleaf skull
2. Parents of normal stature
3. Short-limbs
4. Hypoplastic thorax
5. Polyhydramnios (71% of cases)

f. **Campomelic Dysplasia**
i. Definition: Also called camptomelic dysplasia. A skeletal dysplasia characterized by bent or bowed limbs. Most commonly, the tibia and femurs are affected. It is associated with a wide variety of concomitant anomalies such as congenital heart disease hydronephrosis and hydrocephalus.
ii. Sonographic Findings:
1. Bowing of long bones, especially lower extremity bones.
2. Associated hydronephrosis or hydrocephalus

g. **Short-Rib Polydactyly Syndrome**
   i. Definition: A lethal dysplasia characterized by polydactyly and an extremely narrowed thorax.
   ii. Sonographic Findings:
      1. Polydactyly
      2. Narrowed thorax
      3. Striking micromelia

3. Idiopathic Osteolyses

a. **Pathology**: A group of skeletal dysplasias of unknown etiology that result in diffuse demineralization of bone.

b. **Osteogenesis Imperfecta**
   i. Definition: Disorder of production, secretion or function of collagen. Abnormal fragility of bone caused by hypomineralization. Infants are born with multiple fractures which lead to limb shortening. The skull is soft. Delivery trauma may lead to intracranial hemorrhage and still-birth. No treatment.
   ii. Sonographic Findings:
      1. Presence of fractures or excessive callus formation.
      2. Drastically shortened femur length, bowing.
      3. Hypomineralization of skull

c. **Hypophosphatasia**
   i. Definition: A bony demineralization disorder resulting from low levels of serum and tissue alkaline phosphatase.
   ii. Sonographic Findings:
      1. Short, bowed, demineralized long bones
      2. Marked demineralization of cranium
      3. Increased echogenicity of falx cerebri
      4. Fractures may be present

4. Dysostoses

a. **Pathology**: Absence or malformation of individual bones. Prenatal diagnosis is difficult except in cases of cloverleaf skull (Kleeblattschadel Syndrome). Dolichocephaly and/or brachycephaly may indicate a cranial dysostosis but these conditions may be found as a result of oligo-hydramnios. The cephalic index, (normal 75-85 %) can be useful in making a diagnosis.

b. **Talipes Equinovarus (Club Foot)**
   i. Definition: Can be genetic or environmental. Environmental causes include uterine constraint (oligohydramnios, amniotic band syndrome, uterine tumors). Morphologic anomalies include inversion of the foot and flexion of the sole. The navicular bone deviates closer to the medial calcaneus.

ii. Sonographic Findings:
1. Diagnosis is based on knowledge of the relative orientation of the foot and leg bones.
2. Foot deviated from normal position

c. **Sirenomelia (Mermaid Syndrome)**
i. Definition: Lower extremity fusion which is a severe manifestation of caudal regression syndrome. Morphologic changes include: fusion of lower extremities to varying degrees from membranous attachment of legs to complete fusion of legs with one femur and one tibia.
ii. Sonographic Findings
1. Oligohydramnios
2. Single femur or two femora constantly seen side by side
3. Associated with BRA and multicystic dysplastic kidney

Helpful Hints

While many types and variations of skeletal anomalies have been reported in the ultrasound literature, the major, lethal forms are easily detectable. Grossly abnormal sonographic findings are the hallmark of the conditions incompatible with life. The following table summarizes the unique characteristics of the most common skeletal anomalies.

Anomaly	Unique Characteristic
Achondrogenesis	Absent vertebral ossification
Homozygous achondroplasia	Both parents are dwarves
Heterozygous achondroplasia	Cloverleaf skull Both parents normal stature
Thanatophoric dysplasia	Cloverleaf skull
Campomelic dysplasia	Bowing of tib-fib
Short-rib polydactyly	Small thorax
Osteogenesis imperfecta	Fracture deformities
Hypophosphatasia	“Thin” bones