EMBRYOLOGY

APPENDICULAR SKELETON
Consists of the pectoral and the pelvic girdles and the bones of the limbs. Beginning at the 4th menstrual week primordial bone patterns evolve into cartilaginous bone models (chondrification). By the 6th menstrual week, long bones begin to ossify in the central part of the bone. By 12 menstrual week's primary ossification centers can be identified in nearly all bones of the limbs. Epiphyseal appearance occurs at a specific gestational age in each bone. Sonographic identification of epiphyseal appearance in long bones is a well-established method of determining gestational age. (See Chapter 10: Fetal Biometry)

 TERMINOLOGY

Diaphysis: portion of a bone ossified from a primary center
Epiphysis: portion of a bone ossified from a secondary center.
Epiphyseal cartilage plate: cartilaginous tissue between the diaphysis and the epiphysis

NORMAL SONOGRAPHIC ANATOMY
By 15-16 weeks most bones can be imaged. Ossification center is visualized, not entire structure which contains cartilaginous tissue.

APPENDICULAR SKELETON: imaged well by early-mid second trimester. Long bones are easily seen including metacarpals, metatarsals and phalanges and carpals are not ossified until after birth, therefore they are not seen. Exception is calcaneus which ossifies between 5-6 month. Scapula and clavicle can be seen.
AXIAL SKELETON: cranium, facial bones, pelvis, spine. Sphenoid bone and petrous ridges seen at base of skull separating cranial fossae.
- Orbits, maxilla, mandible and bony nasal septum.
- Pelvis: iliac ossification centers seen from early second trimester. Ischial ossification centers seen at about 20 weeks.
- Spine: can be seen with great clarity especially after 22 weeks. Transverse image offers best method of evaluation. Composed of three ossification centers; two posterior and one anterior. On longitudinal the posterior elements are seen as parallel bands.

CLASSIFICATION OF SKELETAL ANOMALIES:
There are six major categories of skeletal anomalies:

Osteochondrodysplasias
- Defects of growth of tubular bones
- Disorganized development of cartilage and fibrous skeleton
- Abnormalities of density of cortex

Dysostoses
- With cranial and facial involvement
- With predominant axial involvement
- With predominant involvement of extremities

Idiopathic osteolyses
- Osteogenesis imperfecta
- Hypophosphatasia

Miscellaneous disorders with osseous involvement

Chromosomal aberrations

Primary metabolic abnormalities
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 prenatally with ultrasound. Fortunately, the severe conditions that are lethal can usually be detected sonographically. The most common lethal skeletal dysplasias are described below.

**Achondrogenesis**

A rare, lethal form of short-limbed dysplasia which may be inherited genetically. Pathologically it is a failure of ossification process. Two types:

**TYPE I: (Parenti-Fraccaro)**
- Absent vertebral ossification centers
- Incomplete ossification of skull
- Rib fractures
- Arms extremely short and stubby
- Head not enlarged compared to trunk

**Terminology**

<table>
<thead>
<tr>
<th>Term</th>
<th>Definition</th>
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</thead>
<tbody>
<tr>
<td>Rhizomelic</td>
<td>Shortening of the proximal segment of an extremity (humerus, femur)</td>
</tr>
<tr>
<td>Mesomelic</td>
<td>Shortening of the distal segment of an extremity (radius/ulna, tibia/fibula)</td>
</tr>
<tr>
<td>Micromelic</td>
<td>Shortening of both proximal and distal segments</td>
</tr>
<tr>
<td>Amelia</td>
<td>Absence of an extremity</td>
</tr>
<tr>
<td>Polydactyly</td>
<td>Presence of more than five digits</td>
</tr>
<tr>
<td>Syndactyly</td>
<td>Soft tissue or bony fusion of digits</td>
</tr>
</tbody>
</table>

*Illustrated Review of OB/GYN Sonography*
TYPE II: (Langer-Saldino)
- Head large compared to body
- Prominent skin folds over a short neck
- Small chest
- Distended abdomen and possible fetal hydrops
- Very short limbs held away from body

SONOGRAPHIC FINDINGS:
- Lack of vertebral ossification
- Large head with slightly decreased ossification of the cranium
- Severely shortened-limbs (usually involves all limbs)
- Small chest

**Homozygous dominant achondroplasia**
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.

SONOGRAPHIC FINDINGS:
- Both parents are achondroplastic dwarfs
- Cloverleaf skull
- Shortened long bones in 3rd trimester
- Femur/BPD ratio below 1st percentile

**Heterozygous achondroplasia**
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.

SONOGRAPHIC FINDINGS:
- By 27 weeks, femur lengths fall below 99 percent prediction interval
- Rhizomelia
- Normal femur length prior to 20 weeks
**Thanatophoric dysplasia**

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.

**SONOGRAPHIC FINDINGS:**
- *Cloverleaf skull*
- *Parents of normal stature*
- Short-limbs
- Hypoplastic thorax
- Polyhydramnios (71% of cases)

**Campomelic dysplasia**

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.

**SONOGRAPHIC FINDINGS:**
- *Bowing of long bones, especially lower extremity bones*
- Associated hydronephrosis or hydrocephalus

**Short-rib polydactyly syndrome**

A lethal dysplasia characterized by polydactyly and an extremely narrowed thorax.

**SONOGRAPHIC FINDINGS:**
- *Polydactyly*
- Narrowed thorax
- Striking micromelia
A group of skeletal dysplasias of unknown etiology that result in diffuse demineralization of bone.

**Osteogenesis Imperfecta**
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.

**SONOGRAPHIC FINDINGS:**
- Presence of fractures or excessive callus formation
- Drastically shortened femur length and bowing.
- Hypomineralization of skull

**Hypophosphatasia**
A bony demineralization disorder resulting from low levels of serum and tissue alkaline phosphatase.

**SONOGRAPHIC FINDINGS:**
- Short, bowed, demineralized long bones
- Marked demineralization of cranium
- Increased echogenicity of falx cerebri
- Fractures may be present

**Dysostoses**
Absence or malformation of individual bones. Prenatal diagnosis is difficult except in cases of cloverleaf skull (see Kleeblattschadel Syndrome in Other Fetal and Anomalies chapter). Dolichocephaly and/or brachycephaly may indicate a cranial dysostosis but these conditions may be found as a result of oligohydramnios. The cephalic index, (normal 75-85 %) can be useful in making a diagnosis.
**Talipes equinovarus (Club Foot)**
Can be genetic or environmental. Environmental causes include uterine constraint (oligohydramnios, amniotic band syndrome, uterine tumors)

**PATHOLOGY:**
- Inversion of the foot and flexion of the sole
- Navicular bone deviates closer to the medial calcaneus.

**SONOGRAPHIC FINDINGS:**
- Diagnosis is based on knowledge of the relative orientation of the foot and leg bones
- Foot deviated from normal position

**Sirenomelia (Mermaid Syndrome)**
Lower extremity fusion which is a severe manifestation of caudal regression syndrome.

**PATHOLOGY:**
Fusion of lower extremities to varying degrees from membranous attachment of legs to complete fusion of legs with one femur and one tibia.

**SONOGRAPHIC FINDINGS:**
- Single femur or two femora constantly seen side by side
- Oligohydramnios
- 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.

<table>
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<tr>
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<td>“Thin” bones</td>
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