The Fetal Skeletal System

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Ultrasound Assessment of Fetal Skeletal System

- Extremities
- Spine
- Calvarium

Long Bone Development

Ossify by the end of the first trimester
Late third trimester
2° ossification centers visible
distal femur
proximal tibia
proximal humerus

41 weeks

Extremities

Assess
Size
Presence
Bones of forearms
Hand position
Foot position

To Exclude
Skeletal dysplasia
Absent limb
Amniotic band syndrome
Radial hypoplasia
Clenched fists
Clubfoot
Rockerbottom foot
**Femur Length**

Normal for GA = Mean ± 2 SD

- FL falls 2 - 4 SD below mean
  - Most are growth restricted
  - Without skeletal dysplasia

- FL falls > 4 SD below mean
  - Usually a skeletal dysplasia
  - Bones appear abnormal by US

**Long Bones are Too Short**

- Skeletal dysplasia
- Dysotosis
- Malformation
- Deformation
- Amniotic band syndrome
- Restrictive uterine environment
Skeletal Dysplasias

Ultrasound assessment
- Degree of shortening of long bones typically > 4 SD below mean for GA
- Distribution of involved bones: extremities, spine, calvarium, ribs
- Bony abnormalities: ↓ mineralization, fractures, bowing
- Polydactyly

Lethal Skeletal Dysplasias

- Neonate cannot survive usually due to respiratory failure
- Ultrasound diagnosis typically made in 2nd trimester
  - Thanatophoric dysplasia
  - Osteogenesis imperfecta Type 2
  - Achondrogenesis
  - Congenital hypophosphatasia
  - Short rib – polydactyly syndrome

Nonlethal Skeletal Dysplasias

- Infants typically survive
- Ultrasound diagnosis typically not made in 2nd trimester
  - sometimes made in 3rd trimester
  - Heterozygous achondroplasia
  - Osteogenesis imperfecta Types 1&4
  - Asphyxiating thoracic dystrophy

Thanatophoric Dwarf

- Most common lethal skeletal dysplasia
- Severe rhizomelia (proximal shortening)
- Bowel long bones
- Narrowed thorax – short ribs
- Flattened vertebral bodies
- Cloverleaf skull
- Megalencephaly – temporal lobe with excess sulcation/fissures
**Osteogenesis Imperfecta**

**Type 2**
- Type 2 — Autosomal recessive
  - Lethal
- Ultrasound findings — Type 2
  - Fractures
  - Deformities
  - Poor mineralization
  - Soft skull
- Types 1, 3, & 4 — Autosomal dominant
  - Nonlethal

**Osteogenesis Imperfecta Type 2**

**Osteogenesis Imperfecta Type 1 & 4**
- Type 1 & 4 — Autosomal dominant
  - Nonlethal
- Ultrasound findings — Type 1 & 4
  - Lagging growth of long bones
  - 3rd trimester
  - Bowing of long bones
  - Mild deformities
  - Poor mineralization
  - Soft skull

**Osteogenesis Imperfecta Type 1**
Osteogenesis Imperfecta Type 4

Achondrogenesis
Autosomal recessive -- lethal dysplasia
Severely ↓ or absent ossification
Marked limb shortening
Absent vertebral body ossification
Normal calvarial ossification

Arthrogryposis
Multiple joint contractures
Etiologies
- Limitation to movement
- Oligohydramnios
- Multiple gestation
- Bicornuate uterus
- Abnormal nerve function
- Abnormal musculature
- Defective connective tissue
Arthrogryposis Multiplex Congenita

Ultrasound findings
- Contractures
- Intrauterine growth restriction
- Polyhydramnios
- Hydrops

Clenched Hands – Cytomegalovirus
- Ventriculomegaly
- Intracranial calcifications

Arthrogryposis from Larsen syndrome (rare genetic syndrome)
**Abnormal Hands & Forearms**

- Inherently abnormal
- Malformation
- Dysplasia
- Normal tissues effected by external factors
- Deformation
  - uterine constraints
  - amniotic band syndrome
- Disruption (e.g., teratogen)

**Radial Ray Anomalies**

Associated with

- Syndromes
  - Cornelia de Lange
  - Fanconi anemia
  - Holt-Oram
  - Radial aplasia-Thrombocytopenia
  - Poland syndrome
  - Nager acrofacial dysostosis
  - VACTERL
- Trisomies 13 & 18

**Radial Ray Defect**

Abnormal Thumb

VACTERL

**Hypoplastic Radius – VACTERL**

Double-outlet right ventricle & transposition of the great arteries

**Absent radius – Trisomy 18**

**Limb Reduction Defects**

Terminal transverse deletions (e.g., absent hands)

*Isolated*

- sporadic, unilateral
- amniotic band syndrome
- vascular accident

Syndromes

- Orofacial (e.g., Poland)
- Amniotic bands
**Polydactyly**
Supernumerary fingers or toes
Skeletal dysplasias
  - Short-rib polydactyly
  - Chondroectodermal dysplasia
  - Asphyxiating thoracic dysplasia
Trisomy 13
Meckel-Gruber syndrome
Autosomal dominant polydactyly
  usually post-axial

**Clinodactyly & Overlapping Digits**
Deviation or deflection of finger(s)
Curving of 5th finger towards 4th
  Trisomy 21
Overlapping digits
  Trisomy 13
  Trisomy 18

Duplicated thumb

Polydactyly with Meckel-Gruber
Hand
Foot
Clenched hands – Trisomy 18

Clubfoot
Etiology
Genetic
A variety of syndromes
Chromosomal defects
Environmental
Severe oligohydramnios
Uterine anomalies
Ultrasound findings
Bones of the foot lie in parallel to bones of lower leg

Amniotic Band Syndrome
Early rupture of amnion
→ Fibrous bands entrap or adhere to fetus
→ Limb amputations or deformities
  Encephaloceles
  Facial clefts
  Ventral wall defects
  Ectopia cordis

Ultrasound Findings
Fetal deformities
  Skeletal
  Craniofacial
  Ventral wall
  Adherent bands
DDx: Amniotic “sheets”
Spinal Abnormalities

- Meningomyelocele
- Hemivertebra
- Scoliosis
- Diastomatomyelia
- Caudal regression / sacral agenesis
- Sacrococcygeal teratoma

Meningomyelocele

- Normal

Meningocele

Spina bifida
- Protrusion of membranes & fluid
- No protrusion of nerve roots
- Often skin covered
- Ultrasound findings
  - Splaying of posterior elements
  - Cystic mass protruding
Hemivertebrae

Associated with a variety of syndromes
Ultrasound findings
  Kink in spine
  Mismatch of posterior ossification centers

Sacral Agenesis

- Hypoplasia / absence
  2 or more sacral vertebrae
- In fetuses of diabetic mothers with poor glucose control
Cranial Anomalies

- Craniosynostosis
  - Trigonocephaly (Trisomy 13)
  - Cloverleaf skull (Thanatophoric dysplasia)
- Lemon sign (Chiari II malformation)
- Strawberry skull (Trisomy 18)

Craniosynostosis

Premature closure of one or more cranial sutures; Male:Female = 2:1

Complications:
- Abnormal head shape
- Abnormal faces
- Neurologic deficits e.g., hearing loss

Prenatal diagnosis
Typically not possible before 3rd trimester

Cloverleaf Skull

Craniosynostosis causing
trilobed shape
prominent forehead
Associated with
Thanatophoric dysplasia
Cloverleaf skull – Thanatophoric dysplasia

20 weeks

Strawberry-Shaped Cranium

Associated with Trisomy 18

Strawberry-Shaped Cranium
Trisomy 18

16 weeks