The Fetal Skeletal System

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No Disclosures

Ultrasound Assessment of Fetal Skeletal System

- Extremities
- Spine
- Calvarium

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

D: 4.52 cm

No Disclosures
Femur Length
Normal for gestational age
= Mean ± 2 SD
Femur length falls 2 - 4 SD below mean
Most are growth restricted
Without skeletal dysplasia
Femur length 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

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
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

Thanatophoric Dwarf

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

Thanatophoric dysplasia

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
Arthrogryposis

Multiple joint contractures

Etiologies

- Limitation to movement
- Oligohydramnios
- Multiple gestation
- Bicornuate uterus
- Abnormal nerve function
- Abnormal musculature
- Defective connective tissue

Ultrasound findings

- Contractures
- Fetal growth restriction
- Polyhydramnios
- Hydrops

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
Abnormal thumb
VACTERL

Absent radius

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
Nager Acrofacial Dysostosis

Characteristics
- Mandibulofacial dysostosis
- Hypoplastic mandible
- External ear abnormalities
- ± Auditory canal atresia
- Upper extremity reduction defects
- Radial ray defects

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
Polydactyly with Meckel-Gruber

Hand

Foot

Polydactyly

13 weeks

17 weeks

Polydactyly

19 weeks

25 weeks

Ectrodactyly

V-shaped defect (cleft) in middle of hands &/or feet with missing digits

± Syndactyly

Associated

Genetic syndromes, e.g.

Split-hand-foot malformation

Silver-Russell

Cornelia de Lange

Cleft hand
Clinodactyly & Overlapping Digits

Deviation or deflection of finger(s)
Curving of 5th finger towards 4th
Trisomy 21
Overlapping digits
Trisomy 13
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
Rockerbottom Foot

Etiology
- Trisomy 18
- Skeletal dysplasia

Ultrasound findings
- Rounded bottom of foot

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”
Amniotic Band Syndrome

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

13 weeks
21 weeks

Meningomyelocele
Normal
Meningomyelocele

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

Hemivertebrae
Sacral Agenesis

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

Sacral agenesis

Caudal Regression

- Sacral agenesis + Anomalies of pelvis and lower extremities
- ↑ In fetuses of diabetic mothers with poor glucose control

Sacrococcygeal Teratoma

Germ cell tumor arising in presacral area

Ultrasound findings:
- Mass arising from lower sacrum
- Extending posteriorly and inferiorly
- ± Hydrops
- ± Extension anteriorly into pelvis
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

Trigonocephaly
Craniosynostosis with premature fusion of metopic suture
(anterior midline, forehead)
Associated with Trisomy 13

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

Trigonocephaly – Trisomy 13

Semilobar Holoprosencephaly
**Cloverleaf Skull**

Craniosynostosis causing
trilobed shape
prominent forehead
Associated with
Thanatophoric dysplasia

**Lemon-Shaped Cranium**

Associated with
Chiari II malformation
Meningomyelocele

**Strawberry-Shaped Cranium**

Associated with Trisomy 18