

OBSTETRICAL ULTRASOUND SKELETAL DYSPLASIA PROTOCOL (UOBF or UOBC**)

**All exams will be accompanied by either a Detailed Anatomy (UOBC) or Follow Up OB exam (OBF). See separate protocol and image requirements for completion of these exams in addition to below images.

Skeletal dysplasia protocol to be performed when femur or humerus lengths measure **<1st percentile**, or as requested.

***Due to time restraints, patients will be referred to MFM for full survey and additional workup if long bones are incidentally seen on routine imaging. Minimum images to still obtain at routine visit if long bones **measure <2%** are: **Bilateral long bones, chest circumference, sagittal image of chest, detailed anatomy views if only a basic was ordered.**

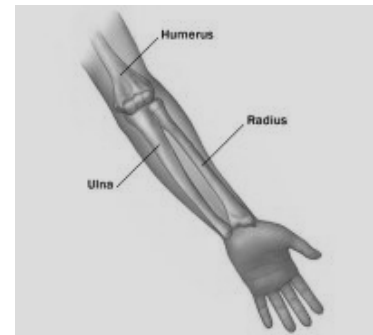
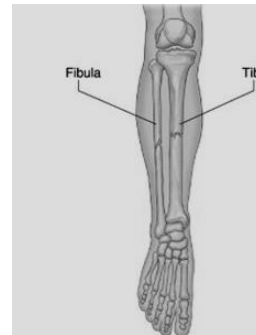
IMAGES TO OBTAIN

EXTREMITIES

1. MEASURE BILATERAL LONG BONES–

Note whether long bones and skull show proper mineralization, curvatures, or fractures:

- Femur
- Tibia
- Fibula
- Humerus
- Radius
- Ulna



2. OBSERVE FOR FETAL MOVEMENT, FLEXION, AND EXTENSION OF LIMBS

3. **FOOT LENGTH** - Measured in coronal plane from the skin over the calcaneum to the first or the second toe. Femur/foot ratio: Normal ≥ 1

4. **CALCANEUS** – Cine sweep through foot, after 24 weeks only

5. **OPEN HAND** showing digit length

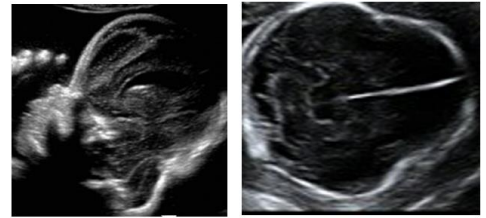


6. EVALUATION OF DIGITS OF THE HANDS AND FEET.

- Polydactyly: presence of more than five digits:
 - Postaxial Polydactyly* -Extra digits are located on the ulnar or fibular side.
 - Preaxial Polydactyly* -Extra digits are located on the radial or tibial side.
- Syndactyly: fusion of adjacent digits
- Clinodactyly: bent or curved deviation of one or more digits.

HEAD

1. **SKULL** - Assess size, shape, and mineralization



Prominent forehead

Cloverleaf skull

2. **ORBITAL DIAMETERS** –

- Inner to inner
- Outer to outer



3. **CEREBELLUM** – Measured in transverse to help verify dating

THORAX

1. **CLAVICLE LENGTH**

2. **SCAPULA** – evaluate if hypoplastic or absent



3. **SAGITTAL AND CORONAL VIEWS OF CHEST** showing chest size in relation to abdomen.

Bell shaped chest



4. **CALCULATE CARDIAC/CHEST CIRCUMFERENCE RATIO:** Chest and cardiac circumferences at the level of the 4-chamber view. Normal ratio is <60%.



5. **RIBS** - Assess size, shape, mineralization, and presence of fractures or fusion of ribs



PELVIS

1. **CORONAL SWEEP THROUGH FETAL PELVIS**

SPINE

1. **ASSESS MINERALIZATION AND CURVATURE.** Use 3D if able

UMBILICAL ARTERY DOPPLERS if indicated for FGR

ADDITIONAL 3D IMAGES for any of concern or structures that cannot be shown well in 2D.

TYPES OF SKELETAL DYSPLASIA

Dysplasia	Prevalence	Etiology	Prognosis	Features
Thanatophoric dysplasia	1 in 10,000	Sporadic	Lethal	Limbs: very short. Thorax: narrow. Trunk: normal. Head: large with prominent forehead.
<i>Type I</i>				Femurs: curved (telephone receiver).
<i>Type II</i>				Femurs: straight. Skull: cloverleaf-shaped.
Osteogenesis imperfecta	1 in 15,000	Autosomal dominant		Fragile bones. Several types but the most severe cases that present prenatally are types II and III.
<i>Type II</i>			Lethal	Limbs: short with fractures. Thorax small with multiple fractures of ribs. Head: hypomineralization of the skull.
<i>Type III</i>			Variable	Multiple fractures, usually present at birth, resulting in scoliosis and very short stature.
Achondroplasia	1 in 25,000	Autosomal dominant	Normal	Limbs: short, but >22 weeks. Head: large with prominent forehead. Spine: lumbar lordosis.
Achondrogenesis	1 in 40,000		Lethal	Limbs: severe shortening. Thorax: narrow. Trunk: short. Head: large with prominent forehead.
<i>Type I</i>		Autosomal recessive		Skull: hypomineralization. Spine: hypomineralization. Thorax: rib fractures.
<i>Type II</i>		Sporadic		Skull: no hypomineralization. Spine: hypomineralization. Thorax: no rib fractures.
Asphyxiating thoracic dystrophy	1 in 70,000	Autosomal recessive	Variable	Limbs: short, but >22 weeks. Thorax: narrow and short.
Ellis-Van Creveld syndrome	1 in 100,000	Autosomal recessive	Variable	Limbs: acromelic and mesomelic shortening, postaxial polydactyly. Thorax: small. Other: heart defects in >50% of cases.
Hypophosphatasia	1 in 100,000	Autosomal recessive	Lethal	Limbs: very short. Thorax: small. Other: hypomineralization of all bones.
Campomelic dysplasia	1 in 200,000	Autosomal recessive	Lethal	Limbs: short, bowed leg bones. Thorax: narrow, hypoplastic scapulae. Head: large with small face.
Jarcho-Levin syndrome	1 in 200,000	Autosomal recessive	Variable	Limbs: normal length. Thorax: short narrow. Trunk: short. Fused vertebral bodies and ribs.
Diastrophic dysplasia	1 in 500,000	Autosomal recessive	Normal	Limbs: very short and bowed. Joints: flexion contractures, talipes. Spine: scoliosis. Other: 'hitchhiker thumb'.

SKELETAL DYSPLASIA IMAGE LIST

IMAGE	MODE
EXTREMITY IMAGES	
BILATERAL LONG BONES	
Femur	2D+
Tibia <i>(medial)</i>	2D+
Fibula <i>(lateral)</i>	2D+
Humerus	2D+
Radius <i>(thumb side)</i>	2D+
Ulna <i>(pinkie side)</i>	2D+
Observe for flexion/extension of limbs	Cine
Foot length	2D+
Calcaneus	Cine
Open hand showing length of digits	2D
Digits of hands	2D
Digits of feet	2D
HEAD IMAGES	
Skull size, shape, and mineralization	2D
Orbits inner to inner	2D+
Orbits outer to outer	2D+
Cerebellum	2D+
THORAX IMAGES	
Clavicle length	2D+
Scapula	2D
Chest and abdomen in sagittal <i>(looking for bell shape)</i>	2D
Chest and abdomen in coronal <i>(looking for bell shape)</i>	2D
Cardiac Circumference	2D+
Thoracic Circumference	2D+
Rib size, shape, and mineralization	2D
PELVIS	
Coronal sweep through pelvis	Cine
SPINE	
Assess mineralization and curvature, 3D if able	2D/3D
UMBILICAL ARTERY	
UA doppler if <10% EFW or AC	2D+
ADDITIONAL 3D IMAGES IF NEEDED	

SKELETAL DYSPLASIA ULTRASOUND PROTOCOL HISTORY

	Date	Changes made	By whom
Created	8/2022	-Separate protocol made - Added to checklist- Sag/Coronal of chest Movement Hands/Feet Digits Ribs Skull UA doppler cerebellum -Chart of SK Types added	Renee Betit Fitzgerald
Added	9/26/2023	Criteria for doing skeletal dysplasia protocol added to intro	Renee Betit Fitzgerald
Added	4/16/2024	Image lists added	Renee Betit Fitzgerald
Reviewed	7/25/2024	Added images: -Scapula -Calcaneus -Open hand showing length of digit -Cine through pelvis -Spine 3D -Additional 3D as needed Workflow change: Due to time restraints, patients will be referred to MFM for full survey and workup if long bones are incidentally seen on routine imaging. Minimum images to still obtain are: Bilateral long bones, chest circumference, sagittal image of chest. Detailed anatomy views if a basic was ordered	Combined Protocol Meeting MFM/RAD Attendees: M. Dighe, E. Cheng, J. Hitti, M. Richley, S Bornemeier, B. Marion, R. Betit Fitzgerald