

# 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 **<1<sup>st</sup> percentile**, or as requested.

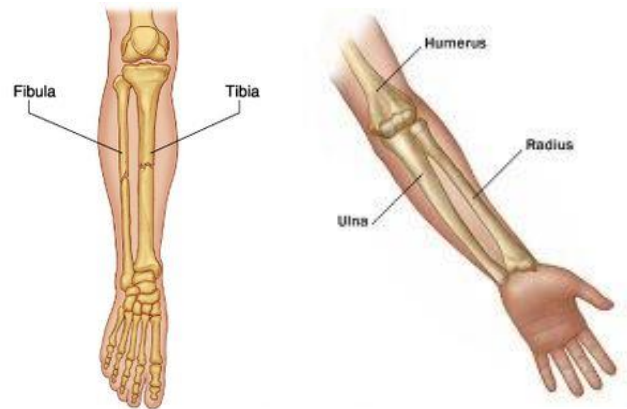
For FL or HL measuring **<2<sup>nd</sup> percentile**, measure all long bones bilaterally and add detailed anatomy views as needed, full skeletal dysplasia protocol not needed.

UW Medicine

## IMAGES TO OBTAIN FOR SKETEAL DYSPLASIA:

1. Measure **bilateral long bones**– Note whether long bones and skull show proper mineralization, curvatures, or fractures:

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



2. Measure the following structures on first visit, not required for follow ups:

- **Foot length** - Measured in coronal plane from the skin over the calcaneum to the first or the second toe. Femur/foot ratio: Normal  $\geq 1$
- **Clavicle length**
- **Orbital diameters** - inner to inner and outer to outer
- **Cerebellum** – Measured in transverse for dating and FGR vs skeletal dysplasia assessment.



3. **Calculate Cardiac/Chest Circumference Ratio:** Normal is <60%. Chest and cardiac circumferences to be obtained at the level of the 4-chamber view of the heart.



4. **Sagittal and coronal view of chest** showing chest size in relation to abdomen.



*Bell-shaped chest*

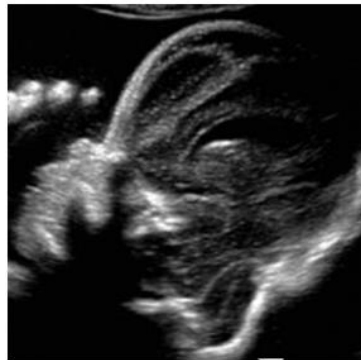


5. **Observe for fetal movement, flexion, and extension of limbs**

6. **Evaluation of hands and feet digits:** several TYPES skeletal dysplasia are associated with alterations 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.

7. **Assess size, shape, and mineralization of skull and head**



*Prominent forehead*



*Cloverleaf skull*

8. **Assess size, shape, mineralization, and presence of fractures in ribs**

9. **Umbilical artery dopplers if indicated**

10. **3D: Only needs to be done to further clarify unanswered questions or if desired by the attending physician**



## TYPES OF SKELETAL DYSPLASIA

Dysplasia	Prevalence	Etiology	Prognosis	Features
<b>Thanatophoric dysplasia</b>	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.
<b>Osteogenesis imperfecta</b>	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.
<b>Achondroplasia</b>	1 in 25,000	Autosomal dominant	Normal	Limbs: short, but >22 weeks. Head: large with prominent forehead. Spine: lumbar lordosis.
<b>Achondrogenesis</b>	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.
<b>Asphyxiating thoracic dystrophy</b>	1 in 70,000	Autosomal recessive	Variable	Limbs: short, but >22 weeks. Thorax: narrow and short.
<b>Ellis–Van Creveld syndrome</b>	1 in 100,000	Autosomal recessive	Variable	Limbs: acromelic and mesomelic shortening, postaxial polydactyly. Thorax: small. Other: heart defects in >50% of cases.
<b>Hypophosphatasia</b>	1 in 100,000	Autosomal recessive	Lethal	Limbs: very short. Thorax: small. Other: hypomineralization of all bones.
<b>Campomelic dysplasia</b>	1 in 200,000	Autosomal recessive	Lethal	Limbs: short, bowed leg bones. Thorax: narrow, hypoplastic scapulae. Head: large with small face.
<b>Jarcho–Levin syndrome</b>	1 in 200,000	Autosomal recessive	Variable	Limbs: normal length. Thorax: short narrow. Trunk: short. Fused vertebral bodies and ribs.
<b>Diastrophic dysplasia</b>	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
<b>BILATERAL LONG BONE MEASUREMENTS</b>	
Femur	2D+
Tibia <i>(medial)</i>	2D+
Fibula <i>(lateral)</i>	2D+
Humerus	2D+
Radius <i>(thumb side)</i>	2D+
Ulna <i>(pinky side)</i>	2D+
<b>OTHER MEASUREMENTS</b>	
Foot	2D+
Clavicle	2D+
Orbits inner to inner	2D+
Orbits outer to outer	2D+
Cerebellum	2D+
Cardiac Circumference	2D+
Thoracic Circumference	2D+
Umbilical Artery if <10% EFW or AC	2D+
<b>OTHER IMAGES TO OBTAIN</b>	
Skull size, shape, and mineralization	2D
Rib size, shape, and mineralization	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
Observe movement & flexion of limbs	2D
Digits of hands	2D
Digits of feet	2D

## 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 -Chart of SK Types added UA doppler cerebellum	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	Renee Betit Fitzgerald