

Approach to the Diagnosis of Skeletal Dysplasias

Skeletal dysplasias are a group of complex and heterogeneous disorders characterized by abnormalities of cartilage and bone. The purpose of this protocol is to:

- 1.) Guide the ultrasound screening and evaluation of a suspected skeletal dysplasia
- 2.) Suggest additional evaluations for a fetus with a suspected skeletal dysplasia
- 3.) Assist clinicians with important elements of patient counseling

A **femur length <5%ile** should prompt:

- Targeted ultrasound
 - Measurement and evaluation of all long bones
 - If isolated short femur, consider re-evaluation in 3-4 weeks

If 1.) other long bones are also <5%ile OR 2.) any long bone is <3%ile OR 3.) other anomalies are identified:

- Additional measures and views (Appendix 1, 2)
- Genetic Counseling Referral
 - Prenatal genetic screening
 - Diagnostic testing
 - Skeletal dysplasia panel
 - Whole exome/whole genome sequencing
 - Single gene NIPT (Baylor PreSeek/Vistara)
- Fetal MRI (*consider to narrow differential or if patient declines genetic counseling*)
- Referral to Pediatric Subspecialties
 - Center for Maternal and Infant Health
 - Neonatal Intensive Care
 - Additional: _____

If the patient elects to terminate pregnancy

- Amniocentesis if not performed yet
- Post-mortem X-Ray and/or MRI (if patient has induction of labor)
- Autopsy (if patient has induction of labor)

If the patient continues the pregnancy

- Antenatal testing
- Growth ultrasounds every 3-4 weeks
- Route of delivery: Cesarean delivery for routine obstetric indications

Appendix 1: Ultrasound Checklist for Suspected Skeletal Dysplasia

PLACENTA

- CERVIX
- PLACENTA
- PCI
- AFI MVP
- Uterus Adnexae

HEAD

- BPD/HC
- Calvarium with compression**
- 3D calvarium if abnormal**
- Brain/Parenchyma
- CSP
- Falx
- Lat Vents (R/L)
- 3rd Ventricle
- 4th Ventricle
- Choroid Plexus
- Cisterna Magna
- Cerebellum
- Nuchal

FACE/NECK

- Profile*
- Nasal Bone
- Face**
- 3D***
- Lenses
- Nose/Lips
- Orbits

SPINE

- Shape and Curvature
- Cervical (Sag/Transverse/Coronal)
- Thoracic(Sag/Transverse/Coronal)
- Lumbar (Sag/Transverse/Coronal)
- Sacral (Sag/Transverse/Coronal)
- 3D Volume**

HEART/CHEST

- 4CH
- w/ color flow
- 3VT 3VV
- LVOT RVOT
- Aortic Arch Ductal Arch
- SVC/IVC
- Ribs (count)**
- Clavicles (presence/absence)**
- Chest circumference**

ABDOMEN/PELVIS

- Visceral Situs
- Lungs
- Diaphragm
- Stomach
- Kidneys
- Abd CI
- Bladder
- 3VC
- Genitalia



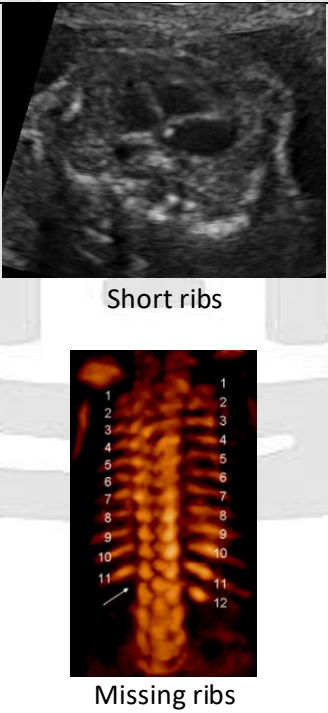
EXTREMITIES

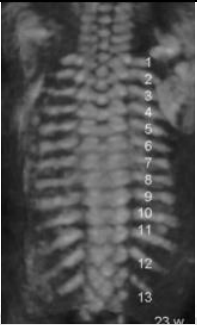
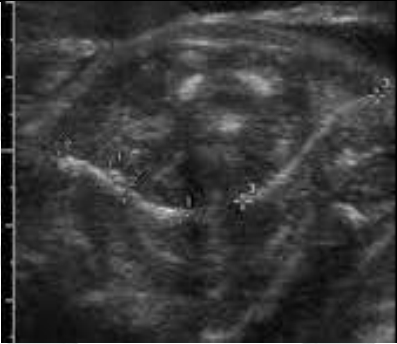
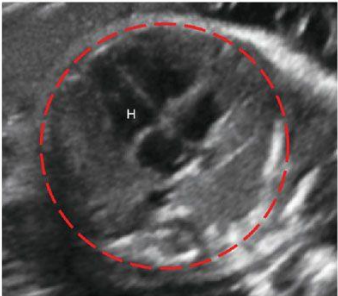
- Humerus
 - Left: Measurement
 - Right: Measurement
- Radius
 - Left: Measurement
 - Right: Measurement
- Ulna
 - Left: Measurement
 - Right: Measurement
- Hand
 - Left: Digit count Architecture/position
 - Right: Digit count Architecture/position
- Upper extremity cine clip for joint mobility**
- Femur
 - Left: Measurement
 - Right: Measurement
- Tibia
 - Left: Measurement
 - Right: Measurement
- Fibula
 - Left: Measurement
 - Right: Measurement
- Foot
 - Left: Digit count Length
 - Right: Digit count Length
- Lower extremity cine clip for joint mobility**

RATIOS

- Chest circumference / abdominal circumference**
- Femur length / foot length**
- Femur length / abdominal circumference**
- Heart circumference / chest circumference**

Appendix 2: Additional Ultrasound Views with Suspected Skeletal Dysplasia

	Image	Notes
<p>Head</p> <p>With and without compression</p>		<p>Distortion with pressure could be evidence of abnormal mineralization (ex: Osteogenesis imperfecta)</p>
<p>Face/Neck</p> <p>Face (with 3D image)</p>		<p>Face should be evaluated for abnormal facies such as hypoplastic/absent nasal bone, clefting, long smooth philtrum, thin upper lip, etc.</p>
<p>Heart/Chest</p> <p>Ribs (size and count)</p>		<p>Should evaluate length, number, shape, fractures, and gaps between ribs.</p>

		
	Extra ribs	
Clavicles		Assess for presence of clavicles, aplasia/hypoplasia.
	Clavicular fracture	
Chest Circumference		Evaluate circumference at level of 4CH view
	Chest circumference	

Appendix 2: Predictors of Life Limiting Pulmonary Hypoplasia

Ratio	Threshold	Studies
CC/AC	<0.6 suggestive of life limiting illness	Yoshimura et al 1996 (Sn: 90.5%; Sp: 90%); Liang et al 2008 (Sn: 50%)
FL/AC	<0.16 suggestive of life limiting illness	Ramus et al 1998 (Sn: 100; Sp: 93%); Rahemtullah et al 1997 (Sn: 100%; Sp: 100%); Nelson et al 2014 (Sn: 91%);
Heart circumference/CC	>0.5 suggestive of life limiting illness	Krakow 2015
Rib size	Encircle <70% of CC at 4CH view (rib cage perimeter/thoracic circumference)	Dugoff et al 1997
Thorax shape	Bell shaped, concave (Coronal view); Narrow AP view	
Thoracic circumference	<5%ile	



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These algorithms are designed to assist the primary care provider in the clinical management of a variety of problems that occur during pregnancy. They should not be interpreted as a standard of care, but instead represent guidelines for management. Variation in practices should take into account such factors as characteristics of the individual patient, health resources, and regional experience with diagnostic and therapeutic modalities.

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