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

<table>
<thead>
<tr>
<th>Image</th>
<th>Notes</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Head</strong></td>
<td></td>
</tr>
<tr>
<td>With and without compression</td>
<td>Distortion with pressure could be evidence of abnormal mineralization (ex: Osteogenesis imperfecta)</td>
</tr>
<tr>
<td>Jeanty 1994</td>
<td></td>
</tr>
<tr>
<td><strong>Face/Neck</strong></td>
<td></td>
</tr>
<tr>
<td>Face (with 3D image)</td>
<td>Face should be evaluated for abnormal facies such as hypoplastic/absent nasal bone, clefting, long smooth philtrum, thin upper lip, etc.</td>
</tr>
<tr>
<td><strong>Heart/Chest</strong></td>
<td></td>
</tr>
<tr>
<td>Ribs (size and count)</td>
<td>Should evaluate length, number, shape, fractures, and gaps between ribs.</td>
</tr>
<tr>
<td>Short ribs</td>
<td></td>
</tr>
<tr>
<td>Missing ribs</td>
<td></td>
</tr>
<tr>
<td>Extra ribs</td>
<td></td>
</tr>
<tr>
<td>-----------</td>
<td></td>
</tr>
<tr>
<td><strong>Clavicles</strong></td>
<td></td>
</tr>
<tr>
<td>Assess for presence of clavicles, aplasia/hypoplasia.</td>
<td></td>
</tr>
<tr>
<td><strong>Clavicular fracture</strong></td>
<td></td>
</tr>
<tr>
<td><strong>Chest Circumference</strong></td>
<td></td>
</tr>
<tr>
<td>Evaluate circumference at level of 4CH view</td>
<td></td>
</tr>
</tbody>
</table>

**Chest circumference**
## Appendix 2: Predictors of Life Limiting Pulmonary Hypoplasia

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


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.

The algorithms remain the intellectual property of the University of North Carolina at Chapel Hill School of Medicine. They cannot be reproduced in whole or in part without the expressed written permission of the school.

www.mombaby.org