Prenatal CT as a tool for dx of severe skeletal abnormalities
PRENATAL W/U FETUS WITH SUSPECTED SD

- Ultrasound
- Echocardiography
- +/- MRI
- +/- fetal CT
- Genetic work up
- Counseling
Ultrasound evaluation

- Femora or humeri < 5th percentile or -2D from mean
- Lethality
  - Small chest circumference
  - Pulmonary hypoplasia
- Lethality by US:
  - Chest/abd wall circumf <0.6
  - Femur length/abd wall circumf <0.16

Table 1 Standardized ultrasound approach to the skeletal dysplasias

- Gestational age based on last menstrual period or first trimester ultrasound
- Length of the long bones (femurs, humerus, radius, ulna, tibia, fibula, and clavicle)
- Shape of long bones (straight, curved, bilateral vs. unilateral)
- Appearance of the metaphyseal ends (spikes, irregularities)
- Echodensity of long bones (well mineralized, poorly mineralized)
- Foot size and shape
- Hands (number of digits, shape of phalanges, mineralization patterns)
- Circumferences (head, abdomen, and chest)
- Lateral view of the chest
- Mineralization and shape of the cranium
- Mineralization and shape of the vertebral bodies
- Size and shape of scapula
- Presence of the secondary epiphyses (calcaneus [>=20 wk] and knee epiphyses [>=28 wk])
- Mandibular size and shape
- Fetal profile (frontal bossing, presence of nasal bone, micrognathia)
- Abnormal posturing of the extremities
- Other congenital anomalies
- Evaluation of amniotic fluid volume (hydramnios)
- Hydrops

Importance of correctly identifying SDs

- Misdiagx can lead to inaccurate recurrence risk info
- Suboptimal management of pts
- Cases should have final dx
  - Autopsy
  - Hystomorphomic analysis of cartilage growth plate
  - Counsel to obtain and storage tissue and/or DNA
- Many skeletal dysplasias associated with significant recurrence risk

- Resources:
  - International Skeletal Dysplasia Registry
  - European Skeletal Dysplasia Network
Low-dose fetal CTs at CHOP

- Started July 2008
- 22 cases to date
- Only fetuses with SEVERE bony abnormality
- Consent
- Only done if dx still in question by US
How to

• CONSENT
• Mark with US the top and bottom of the uterus
• Image ONLY what is strictly necessary
• Topogram
• kVp: 80-100
• mAs: care dose
• Actual scan-few seconds.
• No contrast.

– Then the challenge starts
Radiation load to fetus

- Low-dose fetal CT, NEVER first trimester
- Radiation dose: varies according to maternal size
  - Mean radiation dose: 4.8 mSv
- ACR: 50 mSv is “negligible”
- Houda
  * 100 mSv, threshold for fetal damage
Low-dose Fetal CT dx

- TD: 4
- OI: 2
- IUGR: 2
- Caudal regression syndrome: 2
- Achondroplasia: 1
- Pallister-Killeen: 1
- Cerebrocostomandibular syndrome: 1
- Spondylothoracic dysplasia: 1
- Achondrogenesis: 1
- Mesomelic dysplasia: 1
- Unknown: 3
Fetal CT

- 6 discordant CT and US dx
  - CT correct in 5 cases
  - 1 case, unclear dx

- 5 additional cases, fetal CT contributed to
  - improved visualization of bony anomalies
  - Increased confidence in US dx
CASES
Thanatophoric Dysplasia
Osteogenesis Imperfecta
Mild Oil
Cerebrocostomandibular syndrome
Achondroplasia type II
Spondylothoracic dysostosis
“arthrogryposis”
IUGR
Postmortem

Courtesy Dale Huff MD
postnatal
SUMMARY

• Low-dose fetal CT, complementary imaging tool for evaluation of fetus with SEVERE bony anomalies
• Can improve dx accuracy over US alone
• May facilitate counseling of families

– Work in progress
Thank you!

And thanks to Monica Epelman MD