I have nothing to declare and no financial interest or relationship to disclose.

Take Home Massages...

- Skeletal Anomalies are diverse range of complexities which is NOT Easy to diagnose.
- It is NOT Difficult to detect—just be systematic and gather signs.
- Start with the Femur.
- Signs of poor prognosis (Look at the thorax).
- Multi-team approach.

- Since ancient times, “skeletal deformities” have aroused great fascination that goes far beyond the field of medicine, penetrating into areas such as anthropology and art.
International Nomenclature of Constitutional Disorders of Bones

- The incidence of musculoskeletal anomalies is approximately 0.24%.
- 32 groups.
- 240 disease
ISUOG Practice Guidelines: performance of first-trimester fetal ultrasound scan

### Head
- Cranial bones

### Neck
- Nuchal translucency thickness (if accepted and after informed consent and trained/certified operator available)*

### Spine
- Vertebrae (longitudinal and axial)*

### Extremities
- Four limbs each with three segments
- Hands and feet with normal orientation*

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ISUOG Practice guidelines for performance of the routine mid-trimester fetal ultrasound scan

### Head
- Cranial bones

### Face
- Median facial profile

### Chest
- Normal appearing shape/size of chest

### Skeletal
- No spinal defects or masses (transverse and sagittal views)
- Arms and hands present, normal relationships
- Legs and feet present, normal relationships
The lengths of the humerus, radius/ulna, femur and tibia/fibula are similar and increase linearly with gestation.

18–23-week ➔ the three segments of each extremity should be visualized,

Only necessary to measure the length of one femur.

The relationship of leg and foot should also be assessed to rule out clubfoot.

How Does it Start??

- Index case:
  - Family history of dysplasia,
  - Previous affected babies.

Musculoskeletal Dysplasia

Diagnostic/Clinical Approach...

How Does it Start??

- Incidental finding during routine exam
  - 1st Trimester ➔ High NT, Hydrops, Micromelia.
  - 2nd/3rd Trimester:
    - Micromelia (femur < -4SD),
    - Abnormality of the shape / echogenicity of the bones
    - Abnormal Thorax (Tight, Short...)

Index case:
- Family history of dysplasia,
- Previous affected babies.
**1st Trimester**
- High NT
- Hydrops
- Micromelia

**First clue.**

**Differential diagnosis of a short femur (FL < 2 SD):**
- Normal physiologic variation,
- Intrauterine growth restriction,
- A focal shortening of one femur,
- Abnormal karyotype.

**2nd Trimester**
- The measurement of the femur length (FL) ➝ First clue.
- The differential diagnosis of a short femur (FL < 2 SD):
  - Normal physiologic variation,
  - Intrauterine growth restriction,
  - A focal shortening of one femur,
  - Abnormal karyotype.

**2nd Trimester**
- If the FL is between 2 SD of the mean and 5 mm < 2 SD ➝ interval growth of the FL can be evaluated.
  - During the 2nd trimester the FL normally increases 2.5 mm/week.
- A femur length 5 mm < 2 SD of the mean, a significant skeletal dysplasia is almost certain.

**2nd Trimester**
- The time of onset and degree of FL shortening ➝
  - Heterozygous Achondroplasia ➝ abnormal FL between 21 and 27 weeks.
  - Osteogenesis Imperfecta type II is abnormal at 15 weeks.
**Early onset <20 weeks**
- Thoracic hypoplasia
- Severe limb shortening (micromelia)
- Often Autosomal Recessive (Family history, consanguinity...etc)

**Thoracic hypoplasia**
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**Severe limb shortening (micromelia)**
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**Often Autosomal Recessive (Family history, consanguinity...etc)**

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**Prediction Of Lethality**
- Thoracic circumference <5th percentile.
- Thoracic/abdominal circumference ratio <0.6
- Short thoracic length.
- Ribs that encircle <70% of the thoracic circumference.
- Markedly narrowed anteroposterior (AP) diameter (sagittal view)
- Concave or bell-shaped contour of the thorax (coronal view)
- Femur length/abdominal circumference <0.16

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**Standardized Ultrasound Approach**

To

**The Skeletal Dysplasias**

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**Gestational Age:**

ISUOG-1st Trimester Scan
Length (femur, humerus, radius, ulna, tibia, fibula)

Shape (straight, curved, bilateral vs unilateral)

Echodensity (well mineralized, poorly mineralized)

Abnormal posturing of the extremities
• CHEST AND ABDOMEN:
  - Mineralization and shape of the vertebral bodies

• FETAL PROFILE:
  - TA Ratio >0.8 after 20 weeks

• SPINE:
  - Mineralization and shape of the vertebral bodies

• Other Congenital Anomalies:
  - Evaluation Of Amniotic fluid Volume (Hydramnios)
  - Hydrops
3D Ultrasound

Cranial sutures.

Spine and ribs

Long bones and extremities
Helical CT is a complement to ultrasound, after 26 weeks of gestation it can discriminate the pathological character of a bone abnormality that indicates skeletal dysplasia.

MRI may be somewhat controversial in view of the predominant role of prenatal US.
• The molecular defect has been identified in ≈ 50% of the well-recognized skeletal dysplasias.
• The application of these findings to direct patient care is not yet possible for many of these disorders.
• Long invasive testing to diagnosis interval.