1 **Skeletal Dysplasias**

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2 **Indications**

2 ⦿ Positive family history
- › Prior affected sibling or parent
- › Consanguineous parents

⦿ Appearance of bones on prior study

3 **Extremity measurements**

1 ⦿ Femur
- › Longest dimension
- › Abnormal below 2 standard deviations

⦿ Tibia lateral, Fibula medial

⦿ Humerus
- › Longest dimension

⦿ Ulna-longer proximal to fifth digit distally

⦿ Foot
- ›

2 ⦿ Evaluate for:
- › Bowing angulations
- › Fractures
- › Thickening
- › Decreased mineralization
  - • Absent acoustic shadowing

4 **Extremities: Hands and Feet**

⦿ Club foot (talipes)

⦿ Rocker bottom foot

⦿ Polydactyly

⦿ Sindactyly

⦿ Arthrogryposis (joint contractures)

5 **Head and Face**

1 ⦿ Head
- › Size
- › Shape
  - • Frontal bossing
  - • Cloverleaf
  - • Brachycephaly
  - • craniosynostoses

› Mineralization

2 ⦿ Eyes
› Interorbital distance
› Hyper vs hypotelorism
⦿ Chin
› Micrognathia
⦿ Ears
› Abnormally shaped
⦿

6 **Thorax**

2 ⦿ Thorax
› Measure at 4ch plane
› Length- neck to diaphragm
› Assess ribs
⦿

7 **Spine**

⦿ Relative total length
⦿ Correct curvature
⦿ Mineralization
› Echogenic with acoustic shadowing
⦿ Three ossification centers should be seen
⦿ Platyspondyly
› Flattened vertebral body shape
› Reduced distance between endplates

8 **RADIAL RAY DEFECTS**

❖This is associated with a wide variety of syndromes
❖Absence of the distal radius
❖Radial deviation or clubbed hand
❖Often bowing of the ulna
❖Hypoplastic or absent thumb

9 **Lethal Skeletal Dysplasias**

Severe micromelia
Decreased thoracic circumference
Pulmonary hypoplasia
Associated findings:
Polyhydramnios
Thick redundant skin folds

10 **Thanatophoric Dysplasia**

- Most common lethal dysplasia
- Short extremities
  - Often bowed
- Normal trunk length
- Platyspondyly
- Thorax is narrow
- Large Head-protruding eyes
  - Cloverleaf looking cranium

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1 - 2^{nd} most common
- Severe micromelia
- Macrocranium
- <Thoracic circ.
- <Trunk length
- <Mineralization

12 **Osteogenesis Imperfecta Type II**
- Micromelia
- Decreased thoracic circumference
- Decreased trunk length
- Decreased bone mineralization
  - Absent acoustic shadowing
  - Compression of calvarium
  - See cerebral cortex very well
- Multiple bone fractures
  - Often bowed and angulated

13 **Congenital Hypophosphatasia**

1 - Lethal form of hypophosphatasia
- Deficiency of alkaline phosphatase
- Sonographic:
  - Micromelia
  - Decreased thorax
  - Normal trunck length
  - Decreased mineralization
  - Occasional fractures

14 **Camptomelic Dysplasia**

2 - Bent limb dysplasia
- Short and ventrally bowed tibia and femur
  - May affect upper extremities as well
- Club foot

- Facial abnml
  - Micrognathia and cleft palate

15 **Chondrodysplasia Punctata**

- Craniofacial dysmorphism
- Ocular abnormalities
- Asymmetric limb shortening
- Joint contractures

16 **Short Rib Polydactly Syndrome**

2

- Severe micromelia
- <Thoracic circumference
- Polydactly
- Often have cardiac and GI issues
- Normal-cranial vault, bone mineralization

17 **Nonletal or Variable Prognosis Skeletal Dysplasias**

Heterogenous achondroplasia
Osteogenesis Imperfecta types I, III, and IV
Elleis van Creveld Syndrome
Caudal Regression Syndrome
Vacterl Association
Hand and Foot Syndromes

18 **OI Types I, III, IV-Non Lethal**

- Type I
  - Generalized connective tissue disorder
  - Bone fragility, blue slcera
  - Most fractures from childhood to puberty
- Type II
  - Progressively deforming
  - Often spares humeri, vertebrae, pelvis
- Type IV
  - Mildest form
  - Isolated fractures

19 **Campomelic Dysplasia**

•Shortening and bowing of long bones
•Narrow chest
•Hypoplastic scapula
•Large calvarium
  • Disproportionately small face
•Some males show a female phenotype

20 **Caudal Regression Syndrome**

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- Partial or complete agenesis
  - Lumbar
  - Pelvis
  - Lower limbs
- Associated with maternal diabetes
- Sirenomelia
  - Absent sacrum
  - Fusion of lower ext.
  - Anorectal atresia
  - Renal dysgenesis

21 **Vacterl Association**

2
- Vertebral Defect
- Anal Atresia
- Cardiac Anomalies
- Tracheo-esophageal fistula
- Renal Anomalies
- Limb dysplasia
- Must have at least three

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- Mild to Moderate Micromelia
- Ribs that are short and horizontal
- Polydactly of the hands
- Congenital heart disease
  - Atrial septal defects

23 **Holt Oram Syndrome**

- Congenital heart defect
- Large variety of upper limb anomalies
  - Asymmetrically affected
  - Typically the left affected

24 **ASHPHYXIATING THORACIC DYSPLASIA "JEUNES SYNDROME"**

- ❑Respiratory issues
- ❑Renal dysplasia and cysts
- ❑Small thorax
- ❑Shortened extremities
- ❑Long and abnormally small thorax
- ❑Limbs with variable micromelia

❑Polydactly in about 14%

25 **Amniotic Band Syndrome**

- ⦿ First trimester rupture of the amnion
  - › Amniotic band extending from the chorionic surface to the fetal tissue
- ⦿ May cause
  - › Amputations of limbs/digits
  - › Facial or cranial clefting
  - › Protrusion of uncovered bone
- ⦿ Restriction of motion

26 **Sonographic evaluation**

- ⦿ Limp shortening
- ⦿ Bone contour
- ⦿ Degree of ossificaiton
- ⦿ Thoracic circumference and shape
- ⦿ Hand and foot anomalies
- ⦿ Evaluate fetal face and head
- ⦿ Survey for other anomalies

27 **Summary**

Difficult to diagnose
Require systematic imaging
Often requires multiple disciplines to correctly diagnose
Often fetuses die in utero or are stillborn

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