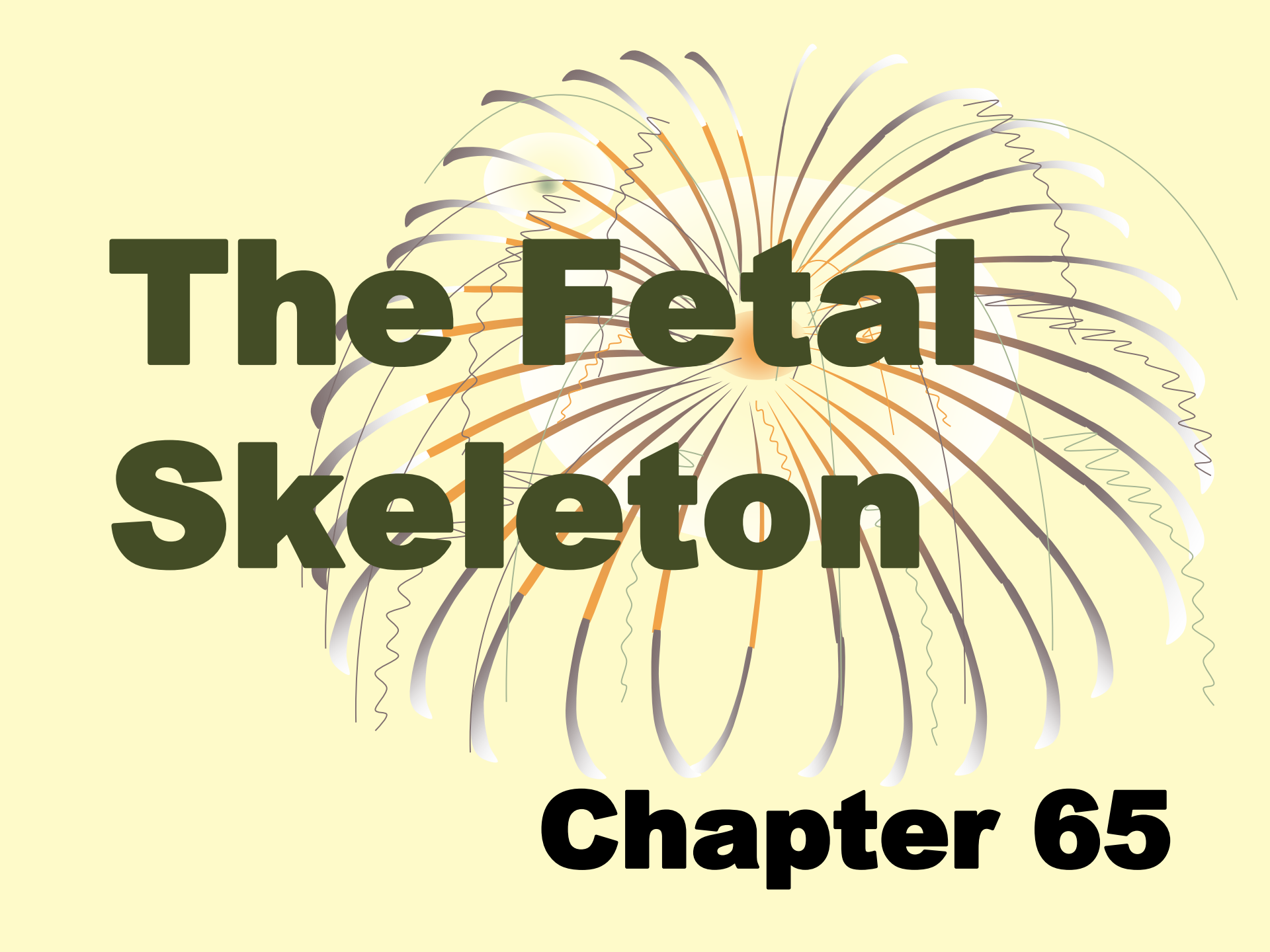


The Fetal Skeleton

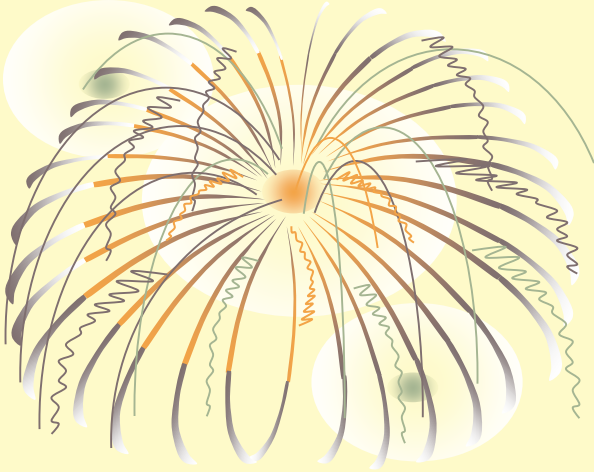
Chapter 65

Embryology

- **Majority of musculoskeletal system forms from**
  - **Primitive mesoderm arising from mesenchymal cells**
    - **Cells arise from different regions of the body**

Embryology

- Limb development begins the 26th or 27th day after conception with the appearance of upper limb buds
- Lower extremity development begins 2 days later
- Fingers are evident by day 49
- Feet and toes essentially complete by the ninth week



Abnormalities of the Skeleton

Skeletal Dysplasia

- Used to describe abnormal growth and density of cartilage and bone
- Dwarfism occurs secondary to a skeletal dysplasia
  - Refers to a disproportionately short stature
- Over 450 types of skeletal dysplasias
  - Not all of seen by sonography

Skeletal Dysplasia

- Some skeletal dysplasias are incompatible with life
- Lethal forms characteristically are extremely severe in their prenatal appearance
- Nonlethal skeletal dysplasias tend to manifest in a milder form

Sonographic Evaluation

- Many skeletal dysplasias are inherited
  - Sporadic occurrences and new mutations do occur
- Majority of skeletal dysplasia will occur in association with
  - Polyhydramnios
  - Other fetal anomalies
  - When risk for recurrence

1. **Assess limb shortening**
   - **All long bones should be measured**
   - **Skeletal dysplasia suspected when limb lengths fall more than two standard deviations below the mean**
2. **Assess bone contour**
   - **Thickness, abnormal bowing or curvature, fractures, and a ribbonlike appearance should be noted**

3. Estimate degree of ossification

- Decreased attenuation of bones with decreased shadowing suggests hypomineralization
- Special attention should be focused toward assessment of the cranium, spine, ribs, and long bones

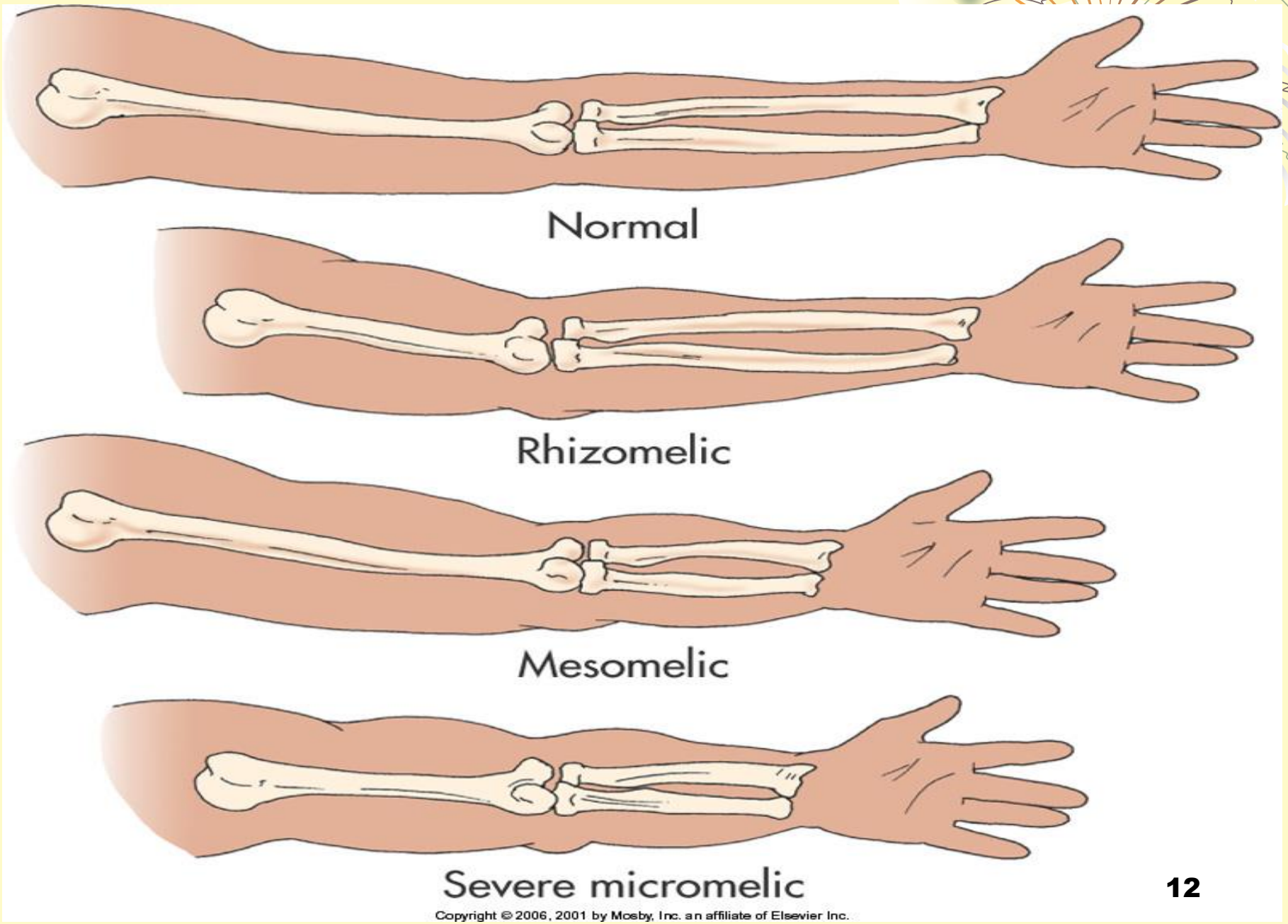
4. Evaluate the thoracic circumference and shape

- Long, narrow chest and a bell-shaped chest may be indicative of specific dysplasias

5. Survey for coexistent hand and foot anomalies
   - Talipes and polydactyly
6. Evaluate face and profile for
   - Facial clefts
   - Frontal bossing
   - Micrognathia
   - Hypertelorism
7. Survey for associated anomalies
   - Hydrocephaly
   - Heart defects
   - Nonimmune hydrops

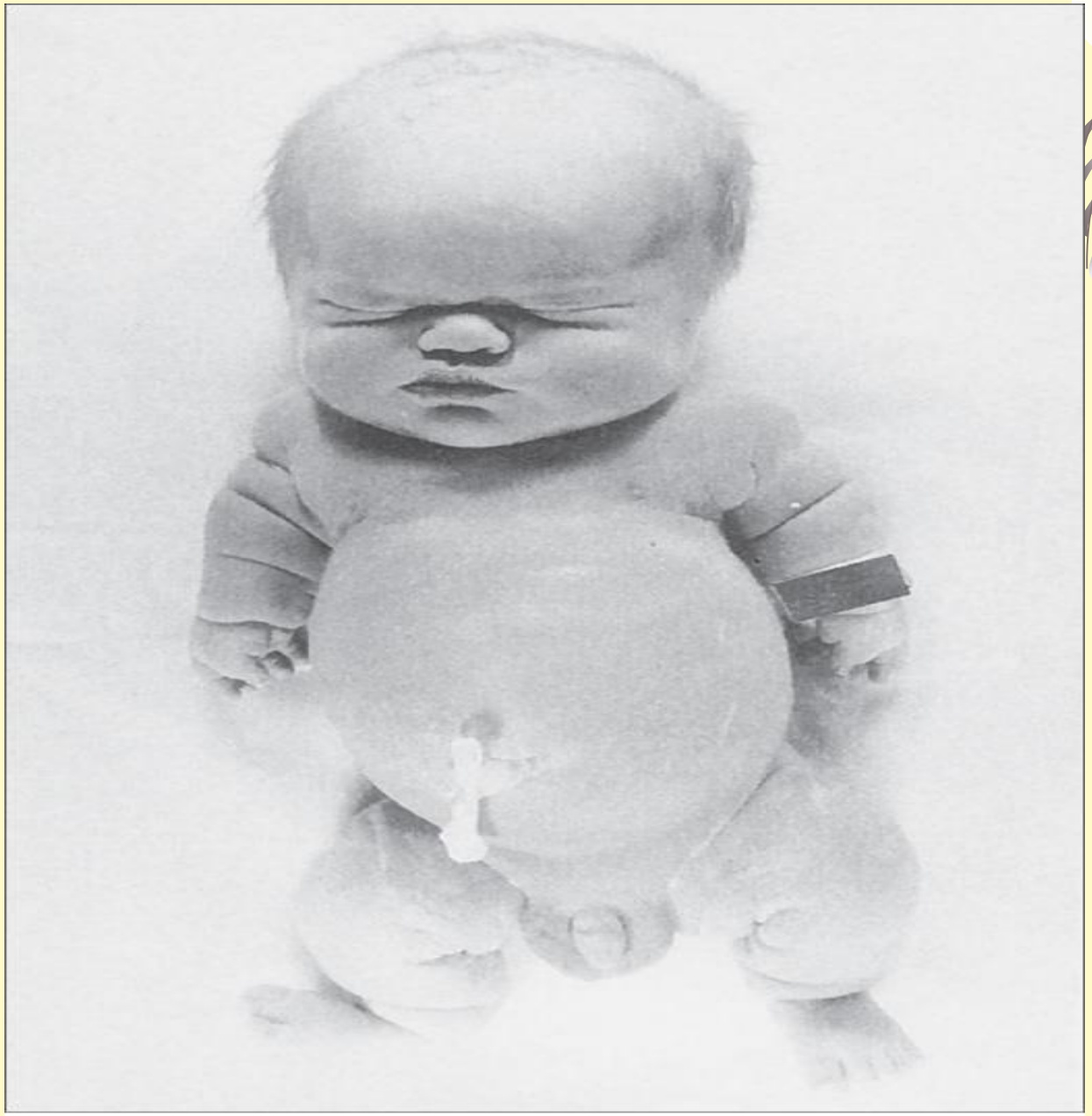
Skeletal Dysplasias

- **Rhizomelia**
  - **Shortening of proximal bones**
    - **Humerus**
    - **Femur**
- **Mesomelia**
  - **Shortening of middle segments**
    - **Radius/ulna**
    - **Tibia/fibula**
- **Micromelia**
  - **Shortening of the entire extremity**



Thanatophoric Dysplasia

- **Most common lethal skeletal dysplasia**
  - **Most infants die shortly after birth**
    - **Pulmonary hypoplasia (narrow thorax)**
- ***Thanatophoros***
  - **Greek word meaning "death personified"**
- **Prognosis is extremely poor**
- **Two main subdivisions**
  - **Type I**
  - **Type II**



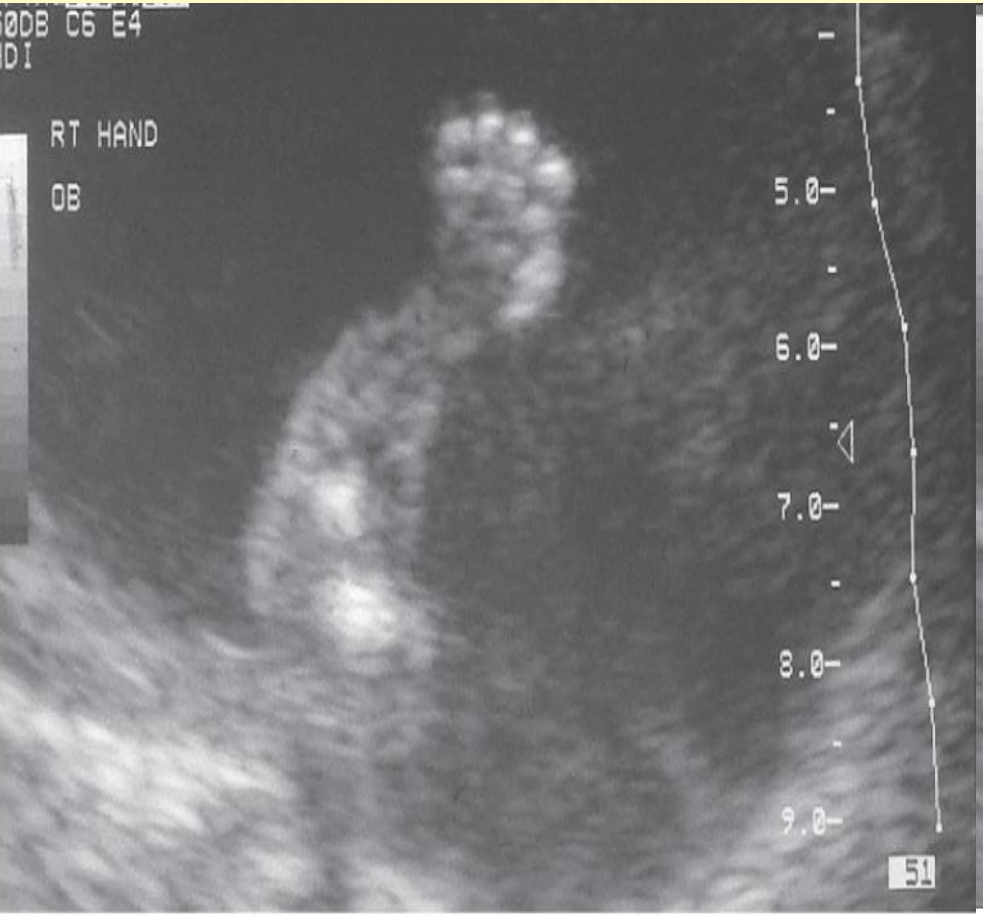
Thanatophoric Dysplasia

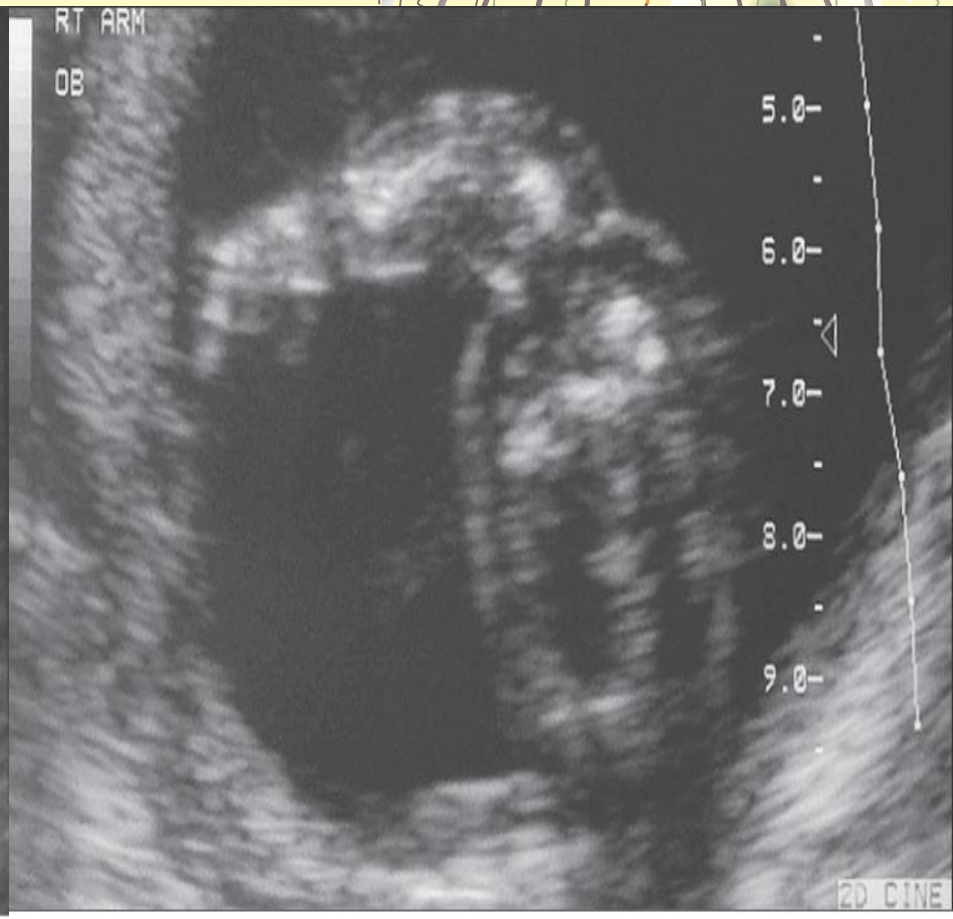
- Type I characterized by
  - Short and curved femurs
  - Flat vertebral bodies
- Type II characterized by
  - Straight, short femurs
  - Flat vertebral bodies
  - Cloverleaf skull
  - May be inherited
    - Autosomal-recessive
- Most cases are sporadic

Sonographic Features

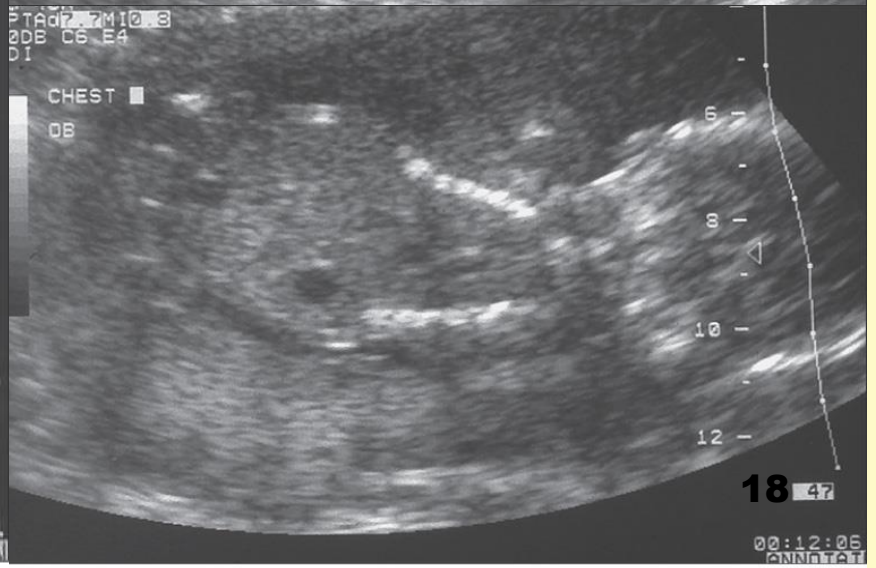
- Severe micromelia
- Narrow thorax with shortened ribs
- Protuberant abdomen
- Frontal bossing (bulging forehead)
- Hypertelorism (widely spaced eyes)
- Flat vertebral bodies (platyspondyly)
- Other sonographic findings may include
  - Severe polyhydramnios
  - Hydrocephalus
  - Nonimmune hydrops

Micromelia

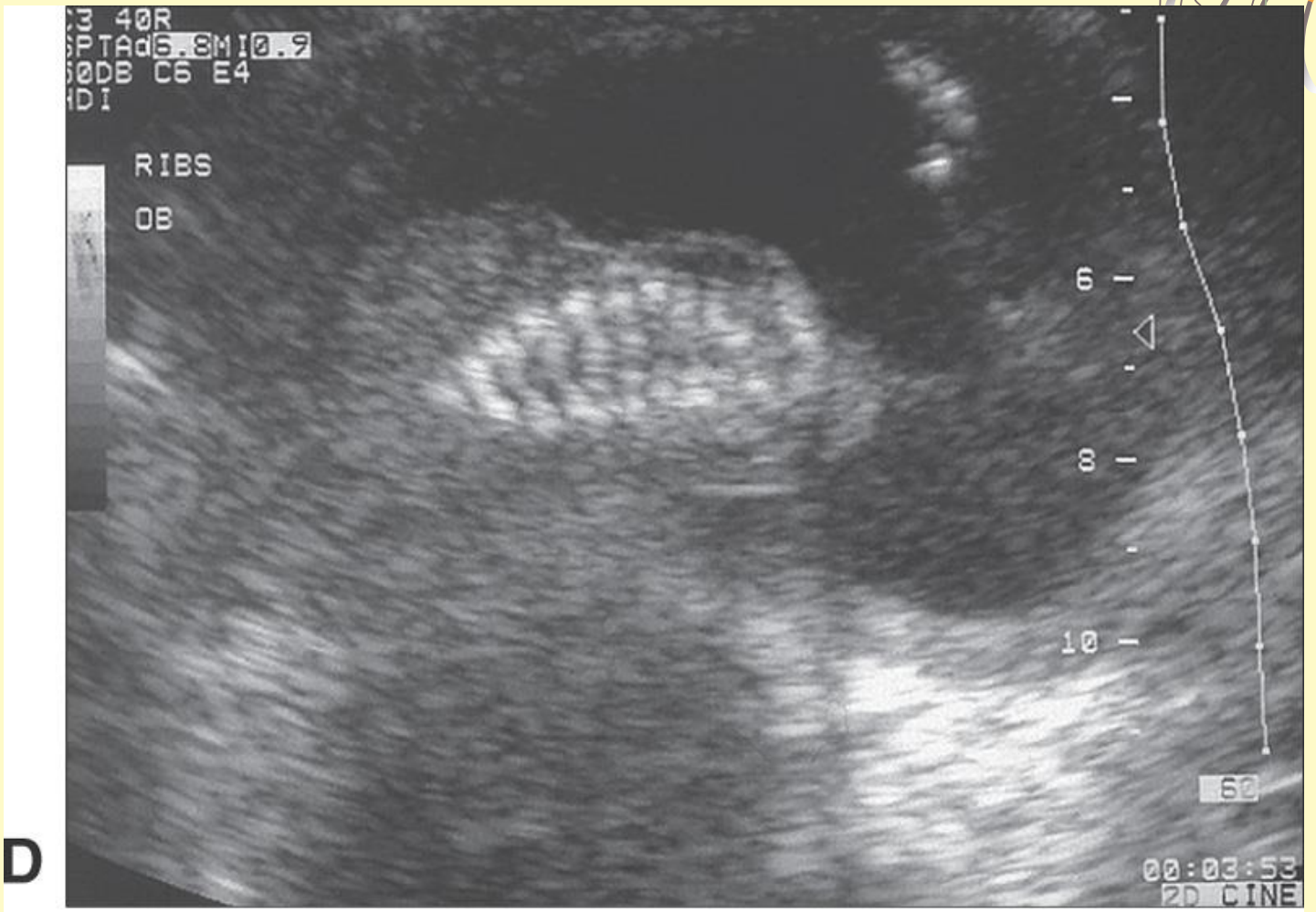




Narrow Thorax



Shortened Ribs



D

Achondroplasia

- Most common nonlethal skeletal dysplasia
- Results from decreased endochondral bone formation
  - Produces short, squat bones
- Sonographic features may not be evident until after 22 weeks
  - When biometry becomes abnormal

Sonographic Features

- Rhizomelia
- Macrocephaly
- Trident hands
  - Short proximal and middle phalanges
- Depressed nasal bridge
- Frontal bossing
- Mild ventriculomegaly
  - (may be identified)

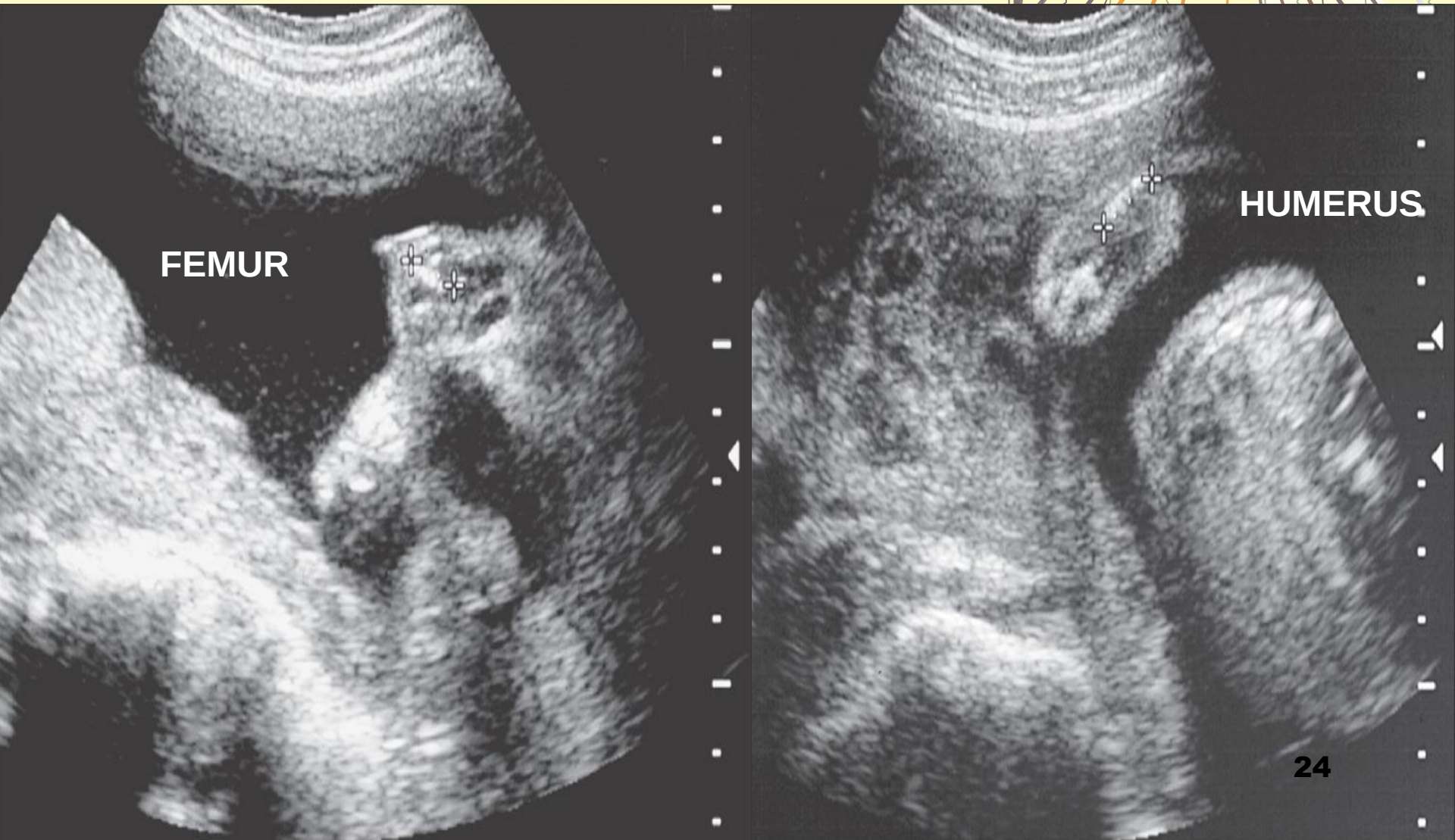
Achondrogenesis

- Rare, lethal skeletal dysplasia
  - Prognosis – poor (pulmonary hypoplasia)
- Caused by cartilage abnormalities
  - Results in abnormal bone formation and hypomineralization
- Two types
  - Type I (Parenti-Fraccaro)
  - Type II (Langer-Saldino)
    - 80% of cases
- Most inherited - autosomal-recessive

Sonographic Features

- Severe micromelia
- Decreased/absent ossification of spine
- Macrocephaly
- Short trunk
- Short thorax and short ribs
- Micrognathia
- Polyhydramnios
- Hydrops (possibly identified)

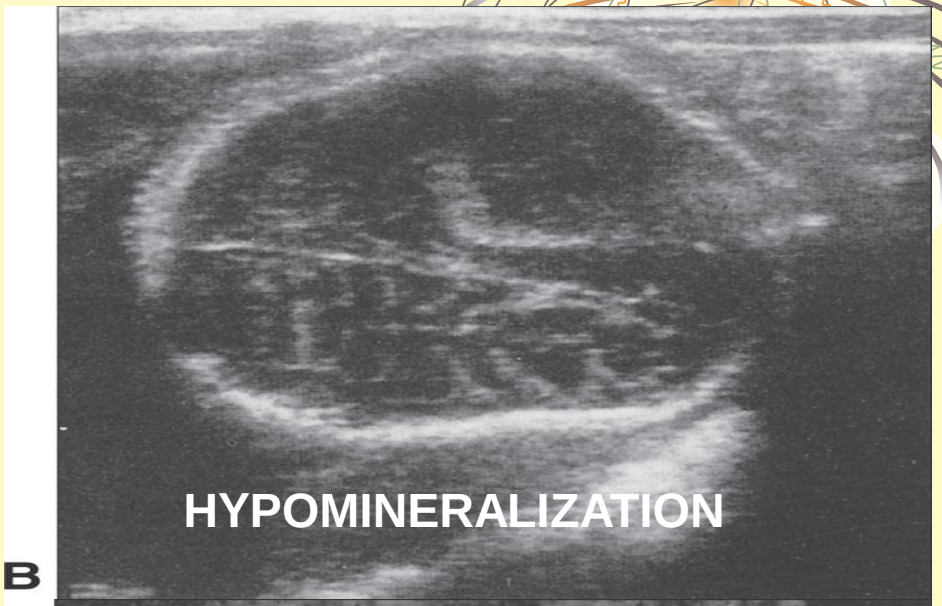
Severe Micromelia

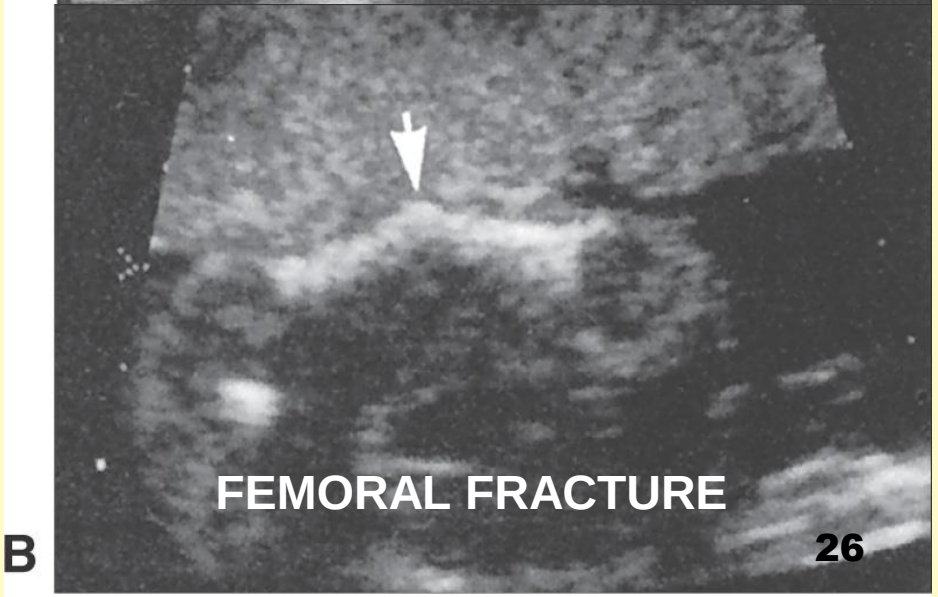


Osteogenesis Imperfecta

- Disorder of collagen production
  - Leading to brittle bones
    - manifestations in
      - Teeth
      - Skin
      - Ligaments
      - Blue sclera
- Four classifications – types I to IV

Osteogenesis Imperfecta





Osteogenesis Imperfecta

- Types I and IV
  - Mildest forms
  - Unlikely differential would be made in utero
- Type III
  - Severe form
- Type II
  - Considered most severe form
    - Lethal outcome

Sonographic Features

- Generalized hypomineralization
  - Especially calvarium
- Multiple fractures of
  - Long bones
  - Ribs
  - Spine
- Narrow thorax
- Micromelia

Congenital Hypophosphatasia

- **Condition presents with diffuse hypomineralization**
  - **caused by an alkaline phosphatase deficiency**
- **Congenital hypophosphatasia is a lethal disorder, with death usually occurring shortly after birth as a result of respiratory complications**

Sonographic Features

- Diffuse hypomineralization
- Moderate to severe micromelia
- Extremities may be
  - Bowed
  - Fractured
  - Absent
- Poorly ossified cranium
  - Well-visualized brain structures
- Small thoracic cavity

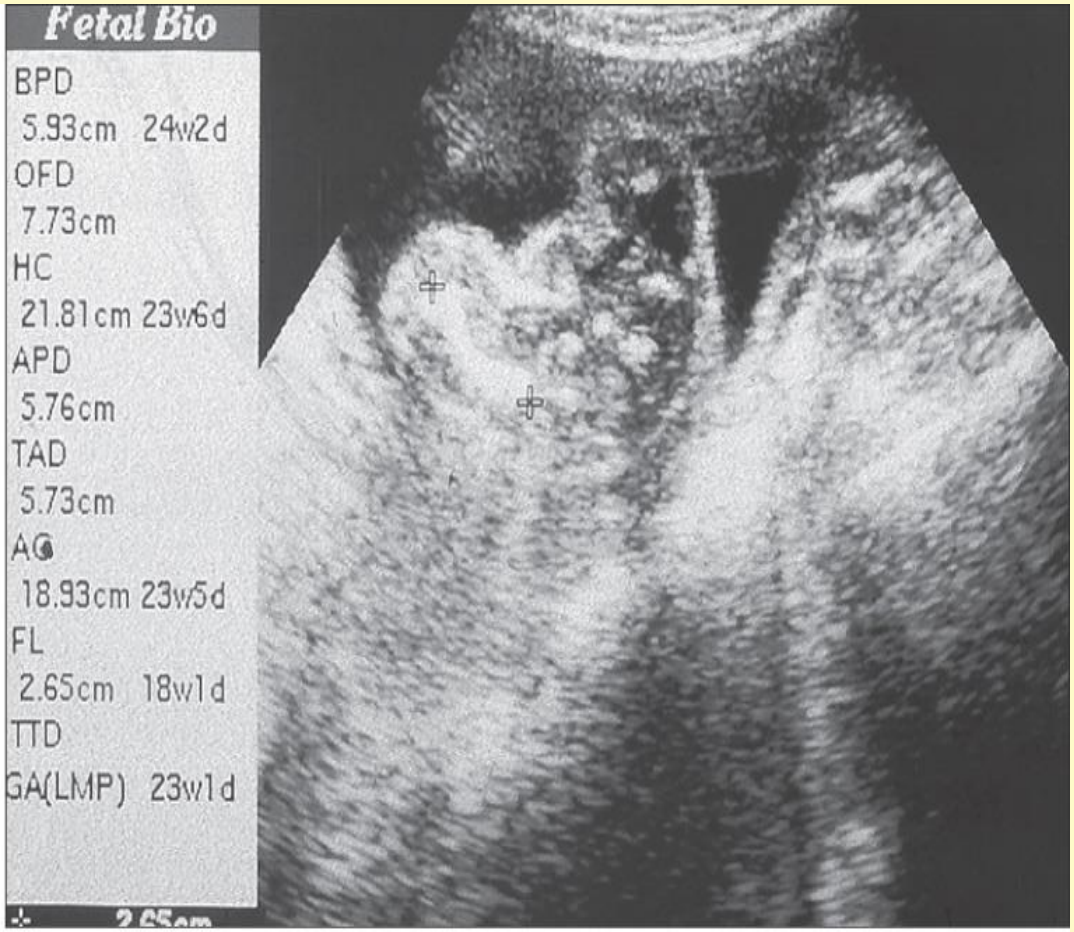
Diastrophic Dysplasia

- Very rare disorder characterized by
  - Micromelia
  - Talipes
  - Cleft palate
  - Micrognathia
  - Scoliosis
  - Short stature
  - Earlobe deformities
  - Hand abnormalities
    - Fixed abducted thumb (hitchhiker thumb)

Diastrophic Dysplasia

(Courtesy Armando Fuentes, MD, Director, Maternal Fetal Center, Orlando, Fla.)

Diastrophic Dysplasia



MICROMELIA

Diastrophic Dysplasia

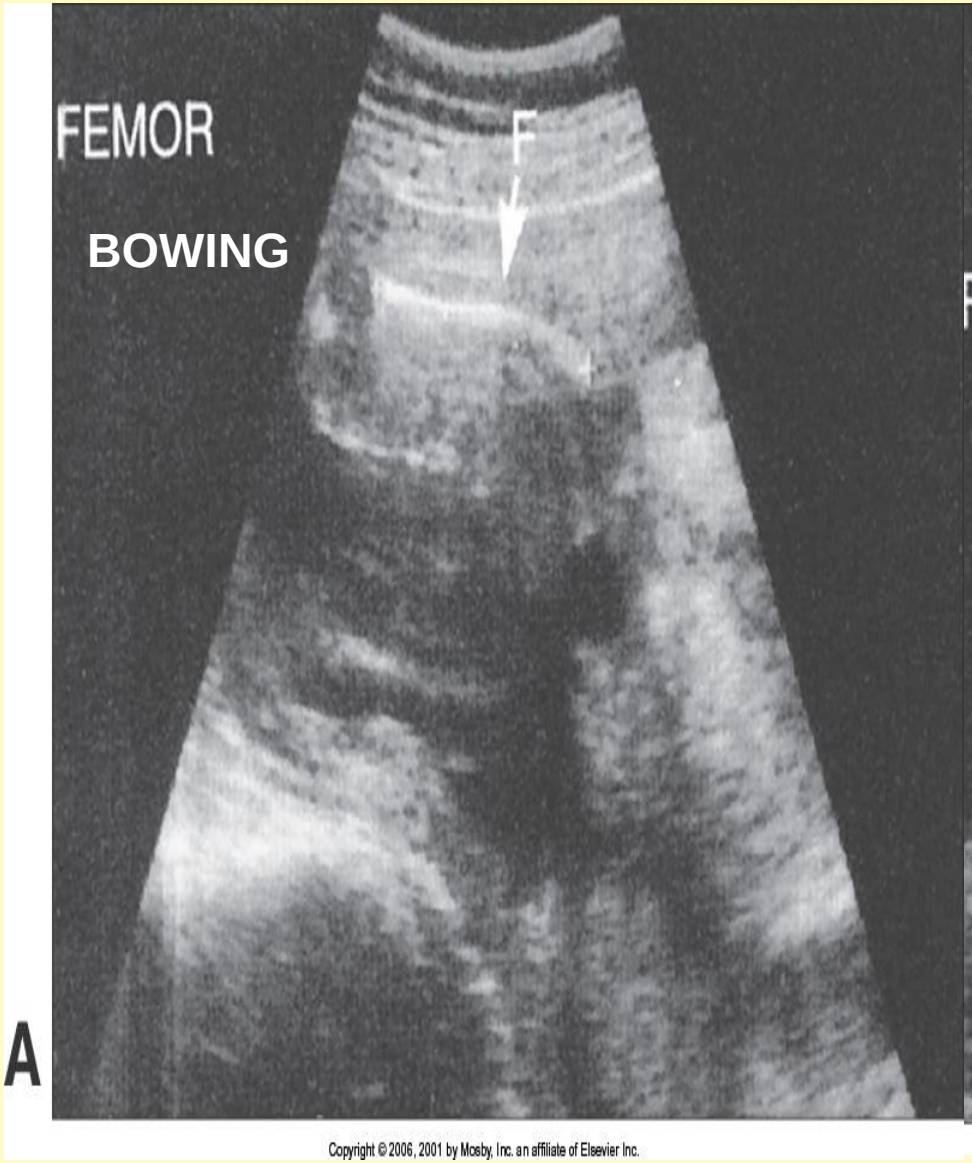
- Prognosis is variable
- Increase in infant mortality
  - Respiratory complications related to
    - Micrognathia
    - Kyphoscoliosis
- Not a lethal disorder
  - Most patients have
    - Normal life span
    - Normal intelligence
- Adult height usually < 4 feet

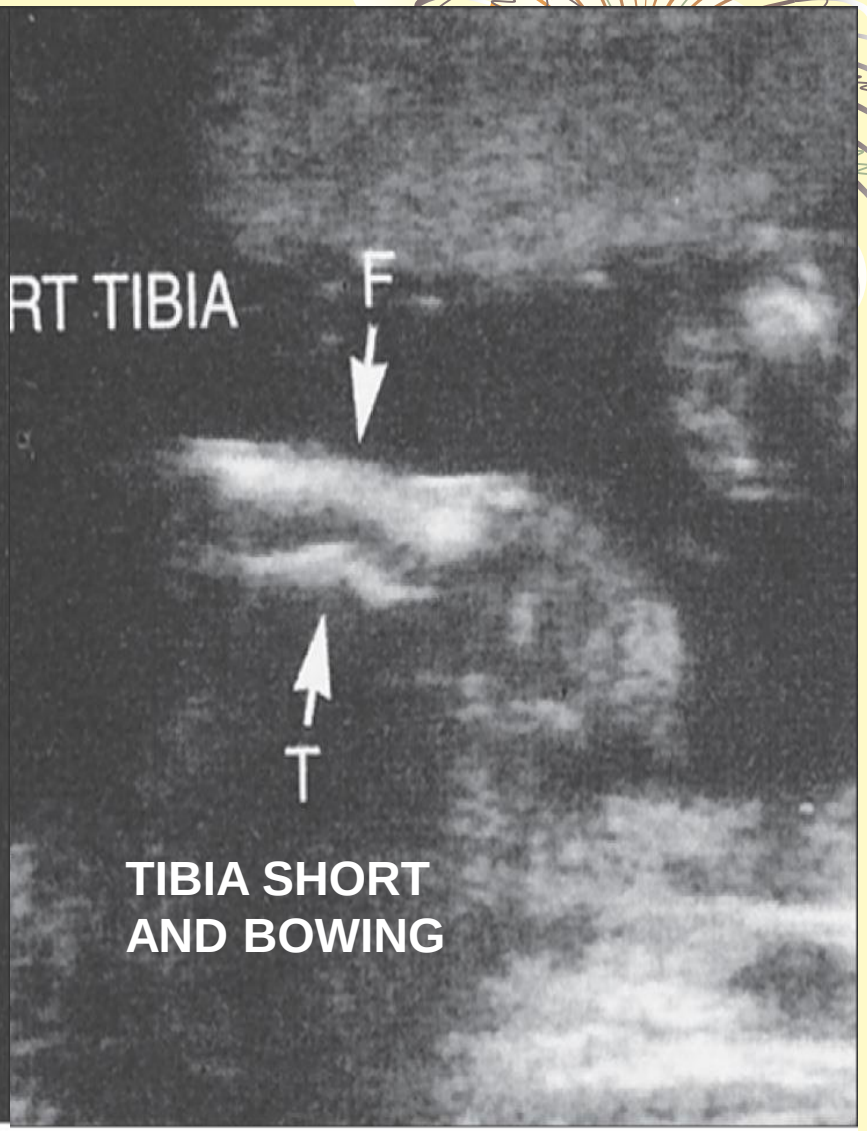
Camptomelic Dysplasia

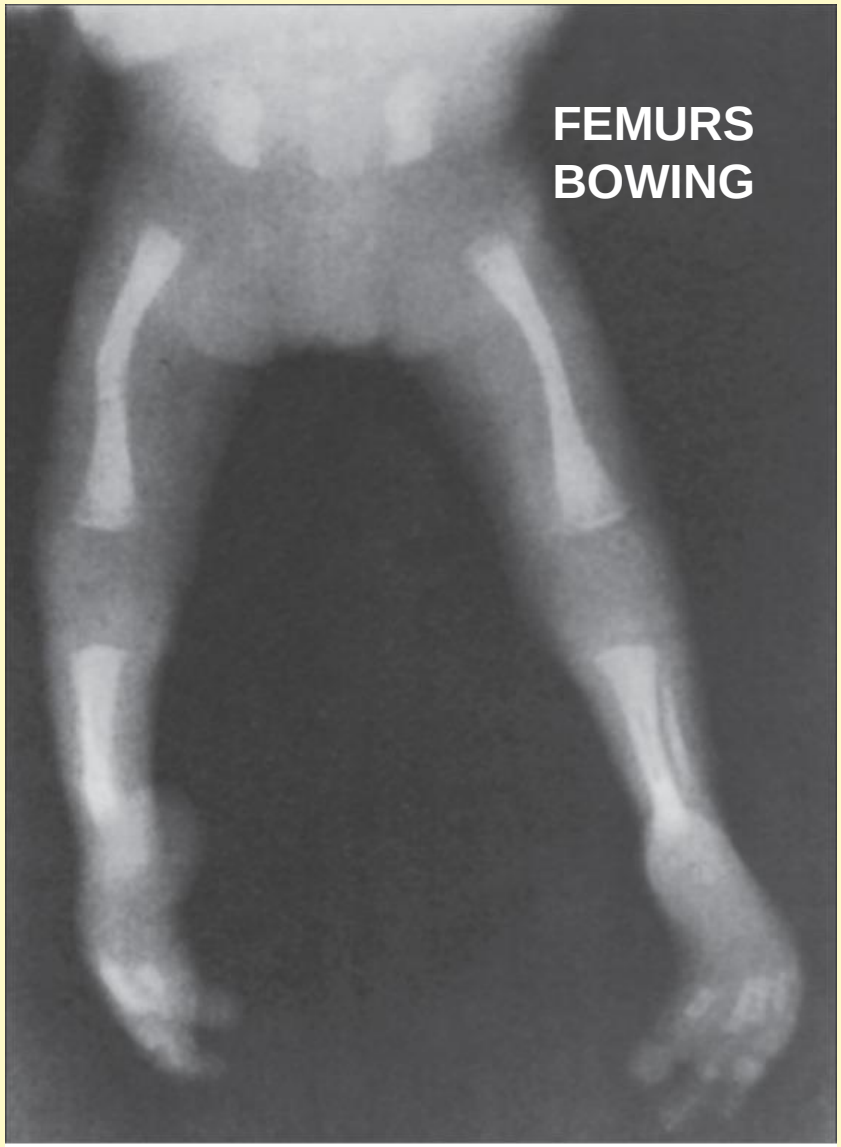
- "bent bone"
  - Group of lethal skeletal dysplasias characterized by
    - Bowing of long bones
- Most cases occur as a spontaneous mutation
- Considered a lethal anomaly
  - Pulmonary hypoplasia

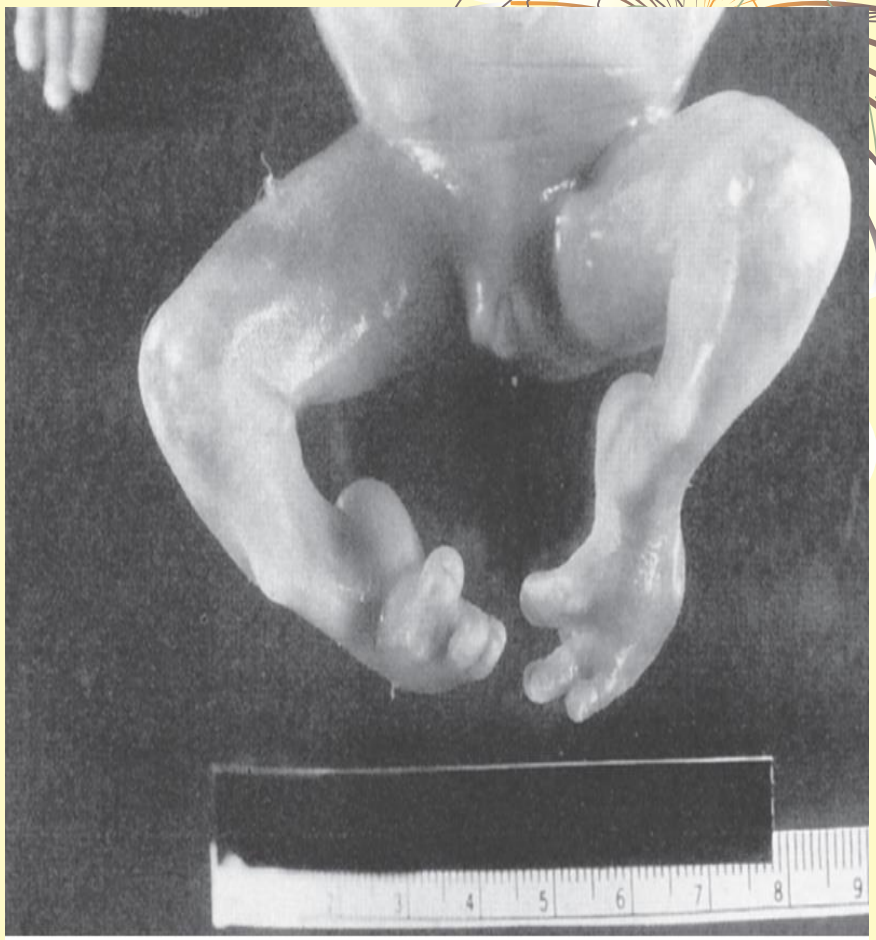
Sonographic Features

- Bowing of long bones
  - lower extremities affected most severely
- Small thorax
- Hypoplastic fibulas
- Hypertelorism
- Cleft palate
- Micrognathia
- Talipes
- Ventriculomegaly
- Hydronephrosis









TIBIAS AND FIBULAS SHORT AND BOWING

Camptomelic Dysplasia

- Infants surviving the neonatal period usually die within the first year
  - Suffer with
    - Respiratory and feeding problems
    - Developmentally delayed
    - Mentally retarded

Roberts' Syndrome

- Rare, characterized by
  - Phocomelia and facial anomalies
- Also known as pseudothalidomide syndrome
- Prognosis is poor
  - Stillbirth and infant mortality are common
- Survivors
  - Growth restricted
  - Severe mental retardation

Sonographic Features

- Phocomelia
  - Upper extremities more severely affected
- Bilateral cleft lip and palate
- Hypertelorism
- Microcephaly
- May be identified
  - Cardiovascular anomalies
  - Renal anomalies
  - Gastrointestinal anomalies

Short-rib Polydactyly Syndrome

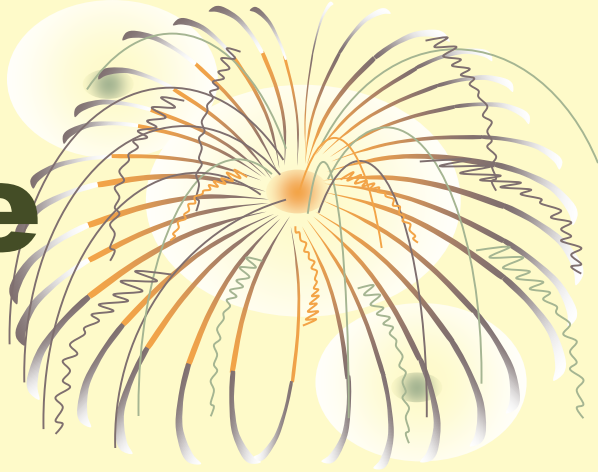
- Lethal skeletal dysplasia
  - Most die shortly after birth
    - Pulmonary hypoplasia
- Characterized by
  - Short ribs and polydactyly
- Four types of dysplasia

Polydactyly

Sonographic Features

- Narrow thorax with short ribs
- Polydactyly
- Micromelia
- Midline facial cleft
- Other sonographic findings include anomalies of the:
  - CNS
  - Cardiovascular system
  - Genitourinary tract
  - Polyhydramnios may also be identified

Jeune's Syndrome

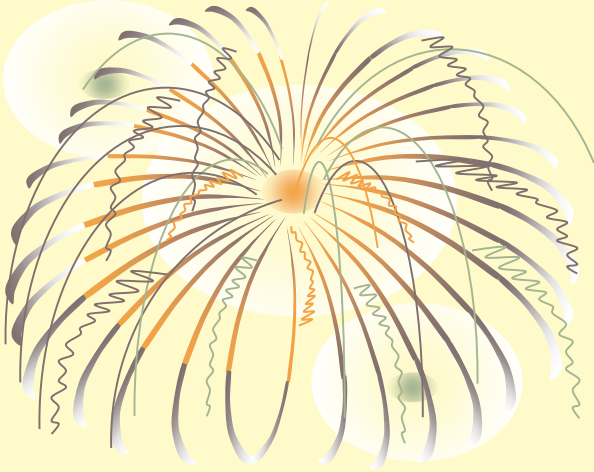


- Also known as
  - Asphyxiating thoracic dysplasia
- Characterized by very narrow thorax
- Range of severity
  - Most severe resulting in death
    - Pulmonary hypoplasia
      - Narrow thorax

Jeune's Syndrome

- **Sonographic features**
  - **Small thorax**
  - **Rhizomelia** **(shortening of humerus/femur)**
  - **Renal dysplasia**
  - **Polydactyly (14%)**

Ellis-van Creveld syndrome



- Also known as
  - Chondroectodermal dysplasia
- Increased frequency in the Amish community
- Survivors are short
  - with normal intellect

Sonographic Features

- Limb shortening
- Polydactyly
- Heart defects (50%)
- May present with
  - Narrow thorax
    - Pulmonary hypoplasia
      - Death in up to one half infants

Caudal Regression Syndrome/Sirenomelia

- **Includes a range of malformations from sacral agenesis to sirenomelia**

  (fusion of the lower extremities)

- **Etiology is not completely understood**
  - **Although it has been associated with diabetes mellitus**

Caudal Regression Syndrome/Sirenomelia

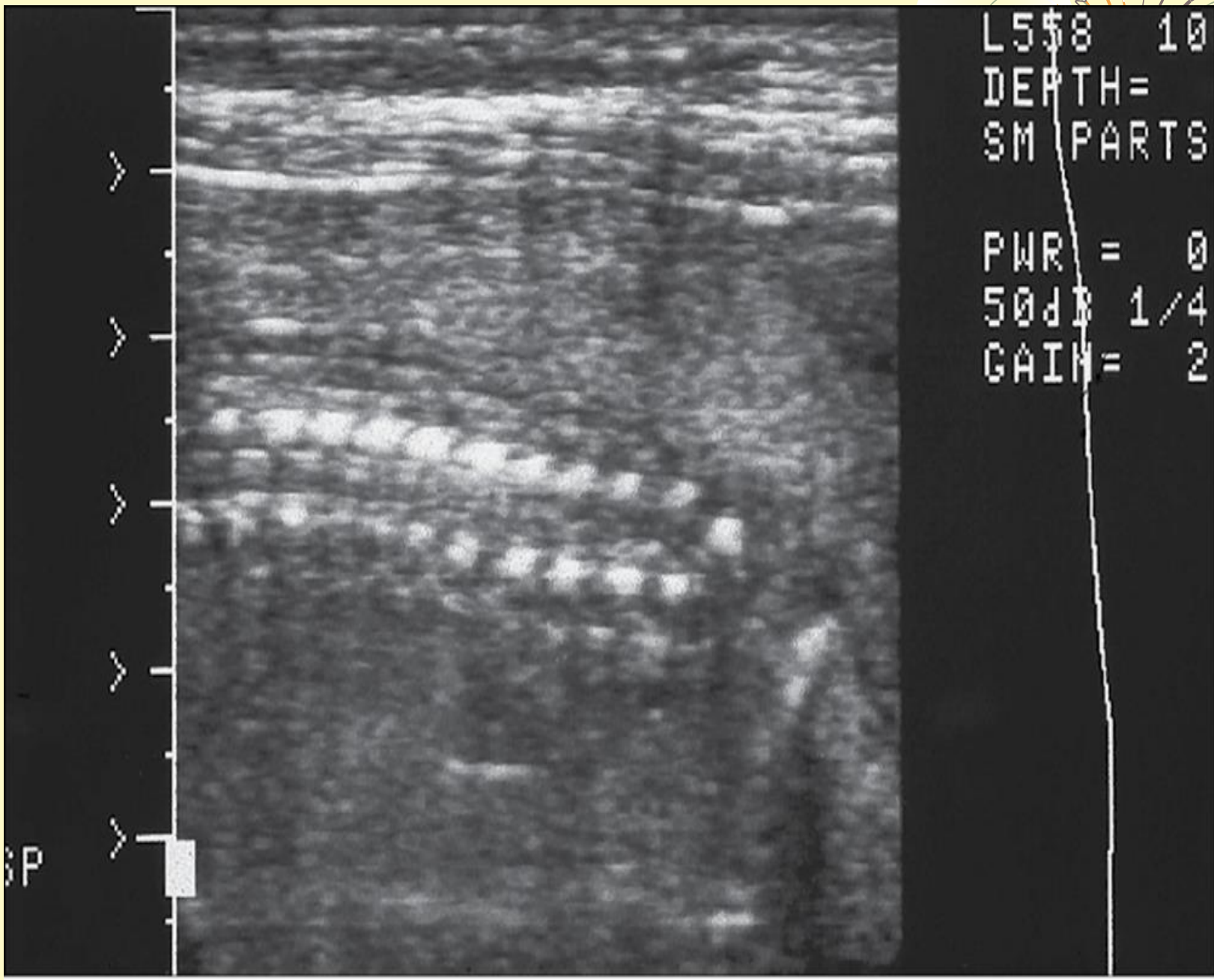
- Genetic factors also have been linked with this disorder
- Vascular hypofusion is thought to be a factor in sirenomelia
  - Single umbilical artery
    - May divert blood flow to the caudal end

Sonographic Features of Caudal Regression Syndrome

- Sacral agenesis
- Talipes
- Abnormal lumbar vertebrae
- Pelvic abnormalities
- May also be seen
  - Contractures or decreased movement of the lower extremities

Sonographic Features of Sirenomelia

- Variable fusion of the lower extremities
- Bilateral renal agenesis
- Oligohydramnios
- Single umbilical artery
- Considered a lethal anomaly
  - Severe renal anomalies results in
    - Oligohydramnios
    - Pulmonary hypoplasia



(Courtesy Armando Fuentes, MD, Director, Maternal Fetal Center, Orlando, Fla.)

Other Associated Anomalies

- **Anal atresia**
- **Heart defects**
- **Gastrointestinal**
- **Genitourinary**
- **Prognosis for CR depends on the severity and the associated anomalies**

VACTERL Association

- Group of anomalies that may occur together or in combination with each other
  - Vertebral defects
  - Anal atresia
  - Cardiac anomalies
  - Tracheoesophageal fistula
  - Renal anomalies
  - Limb dysplasia

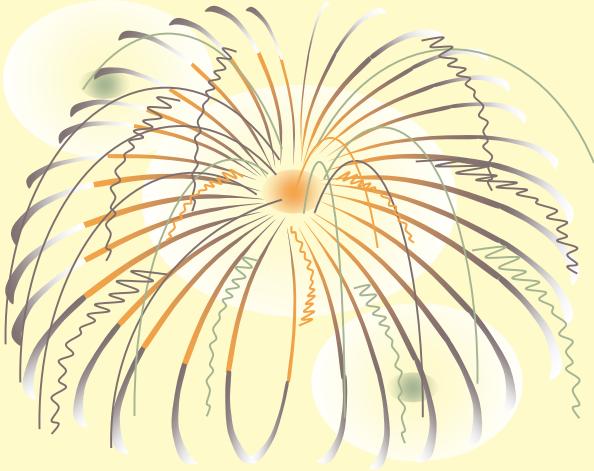
VACTERL Association

- For VACTERL association to be considered
  - Three features must be identified
  - Single umbilical artery has been noted in association with VACTERL

Miscellaneous Limb Abnormalities

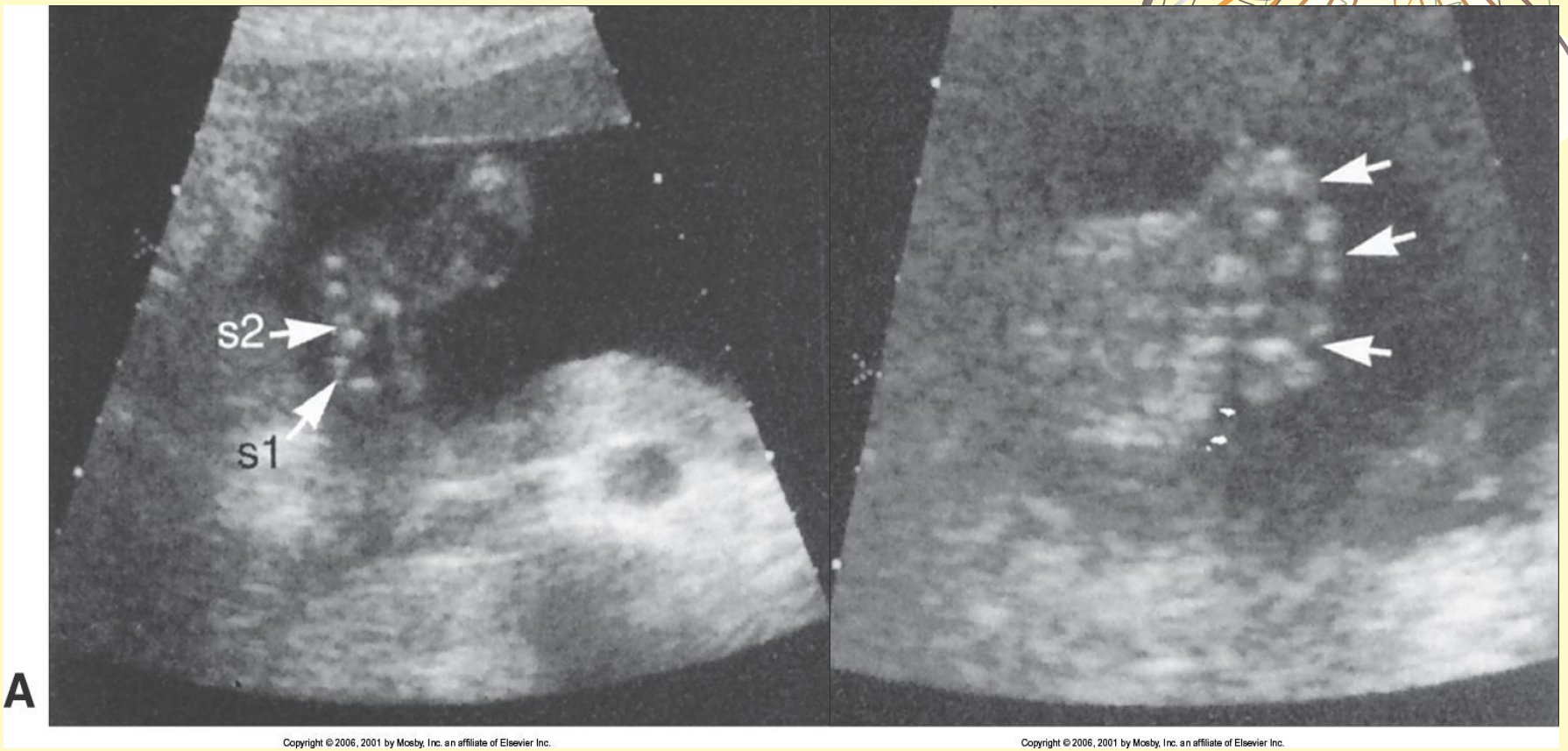
- Hand and foot abnormalities may occur as
  - Part of a chromosomal syndrome
  - an isolated event
- Amputation defects
  - Amniotic band syndrome
- Congenital absence of one or more extremities (amelia) may be observed prenatally

Hand Anomalies



- Missing digits
- Fused digits (syndactyly)
- Split hand (ectrodactyly)
  - Lobster-claw deformity
- Extra digits (polydactyly) may be
  - Isolated
  - Part of a syndrome
  - Chromosomal anomaly
- Overlapping digits (clinodactyly)

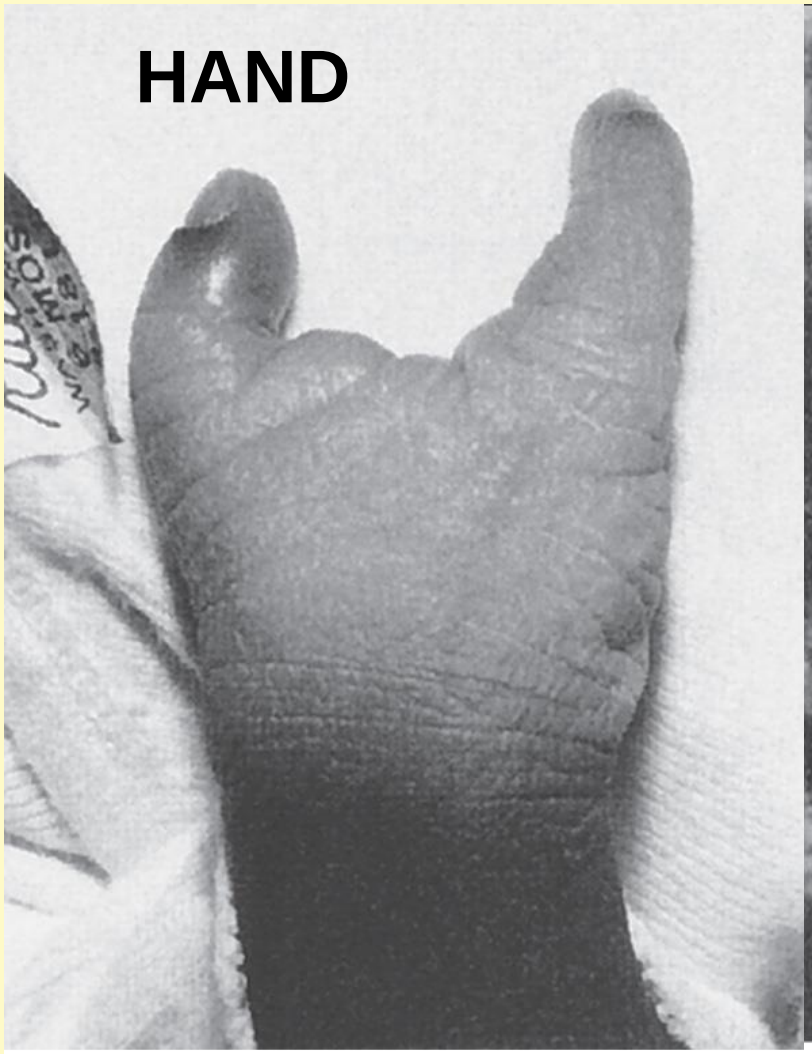
Polysyndactyly

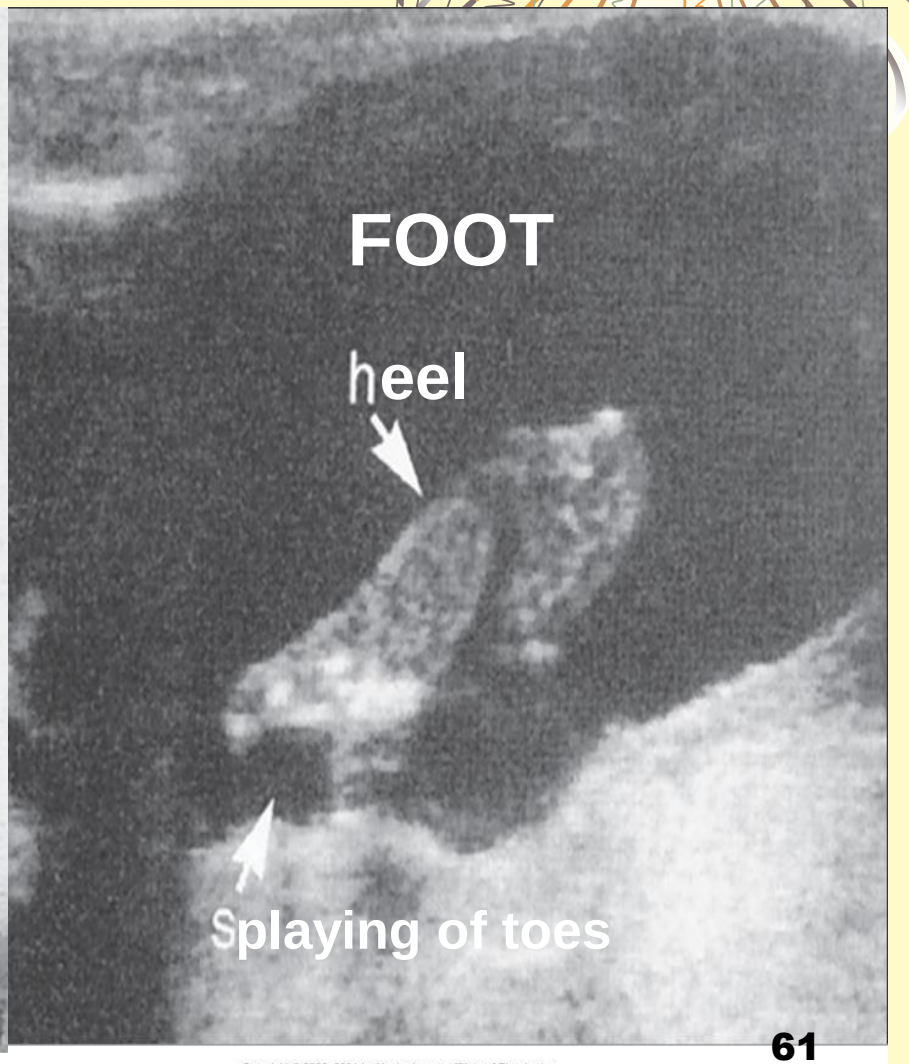


Webbing (syndactyly) of the first and second toes *(s1)* and of the third and fourth toes *(s2)*
Six toes were present (polydactyly)

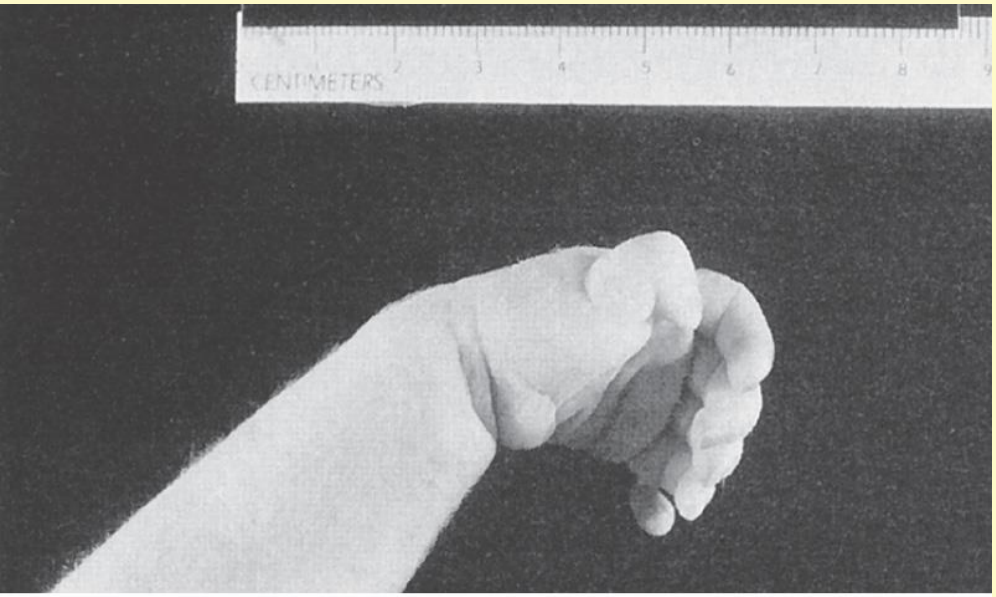
Malalignment of phalanges

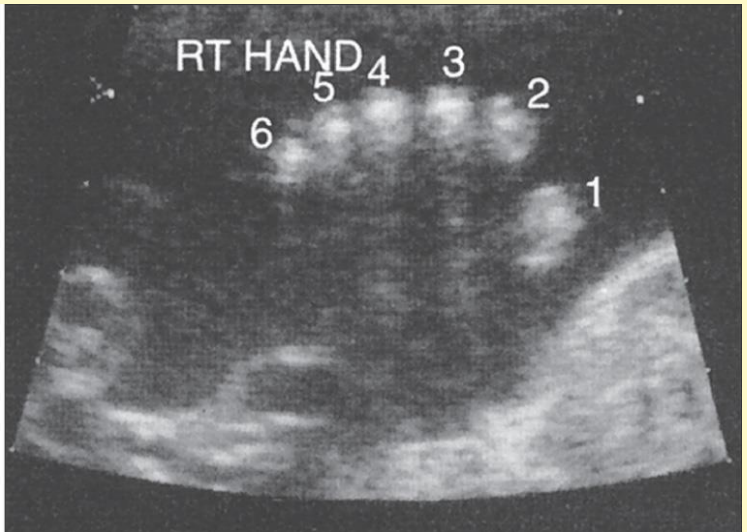
Ectrodactyly



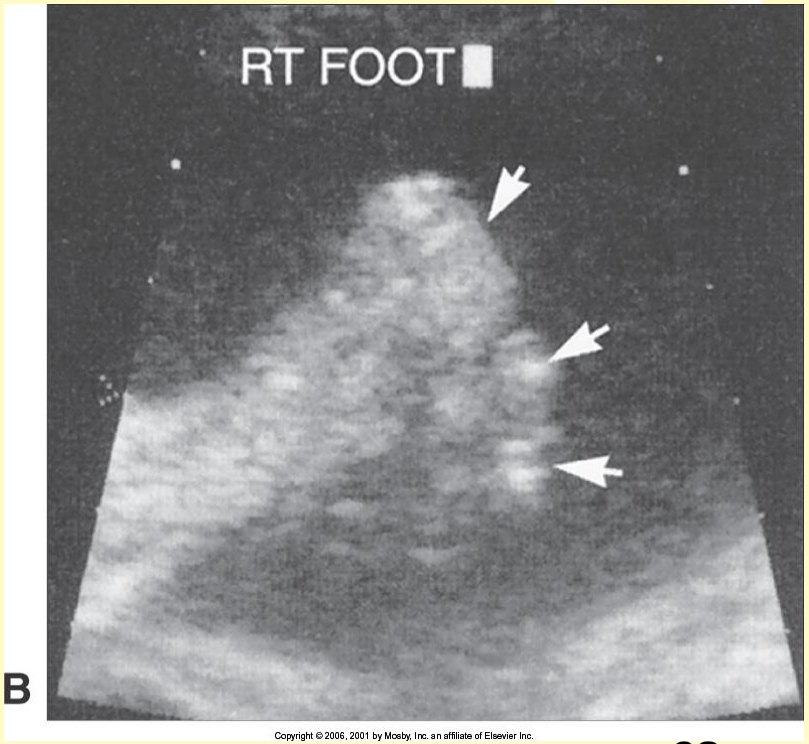


Polydactyly

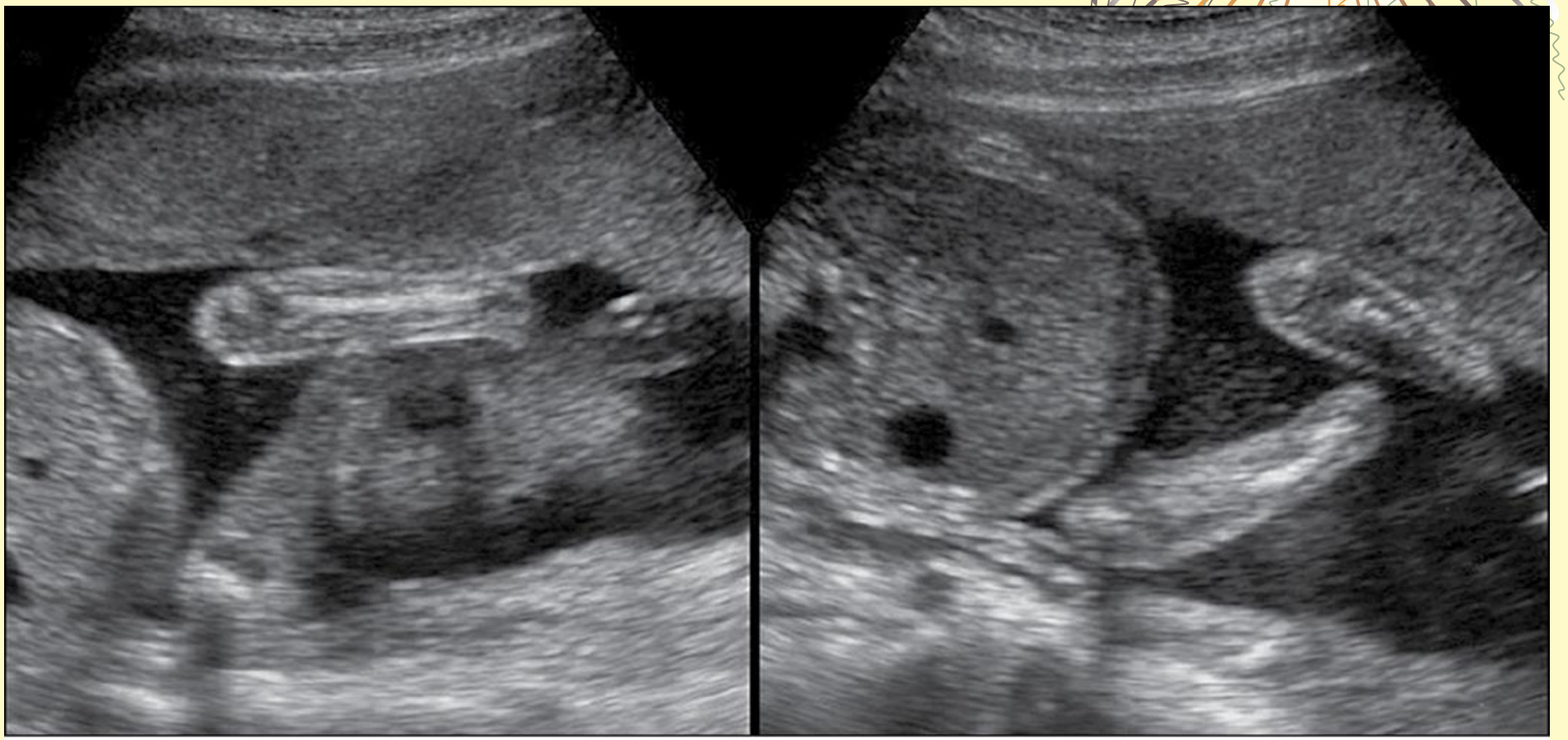




DUPLICATION OF GREAT TOE



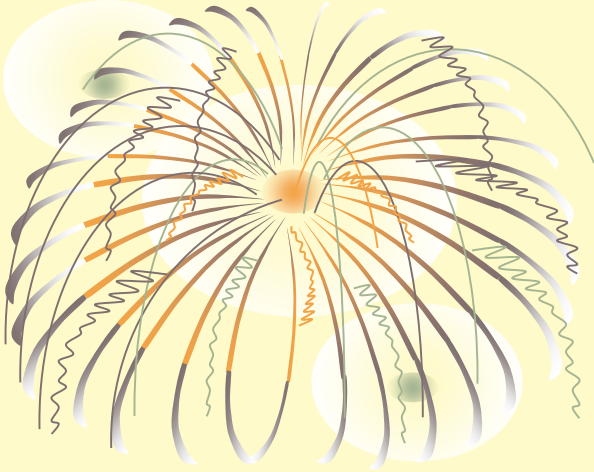
Achieropodia



A

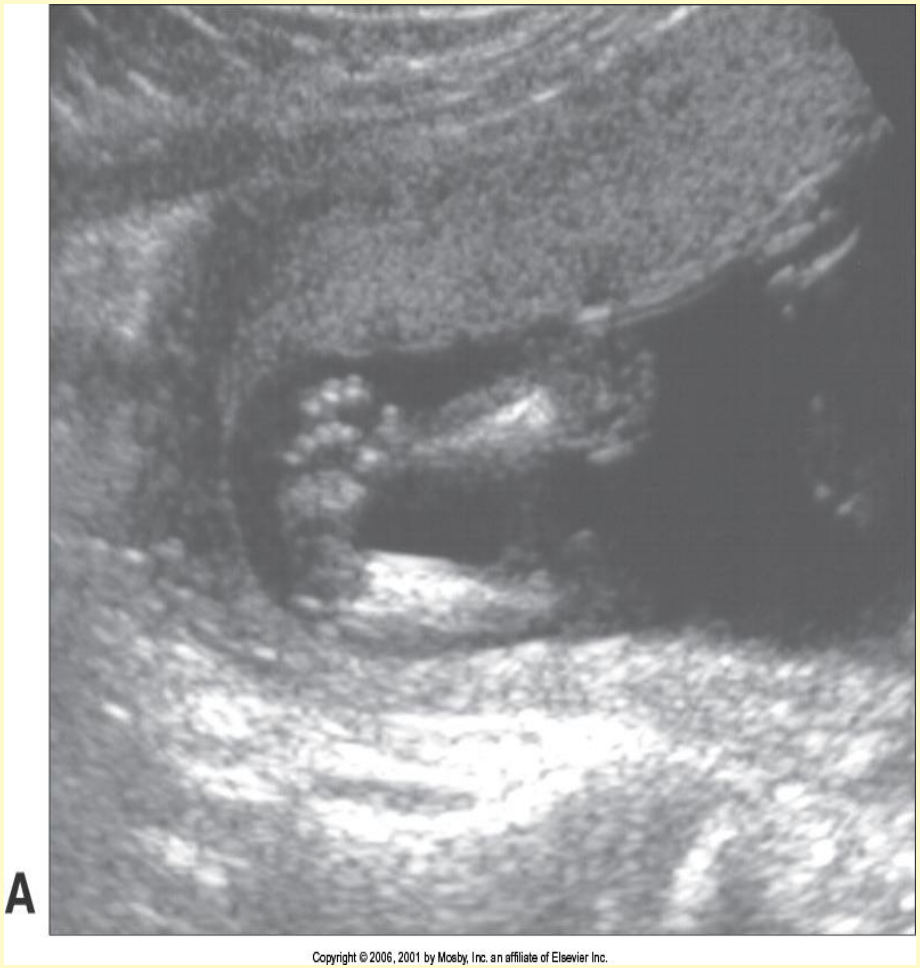
Symmetric absence of hands and feet

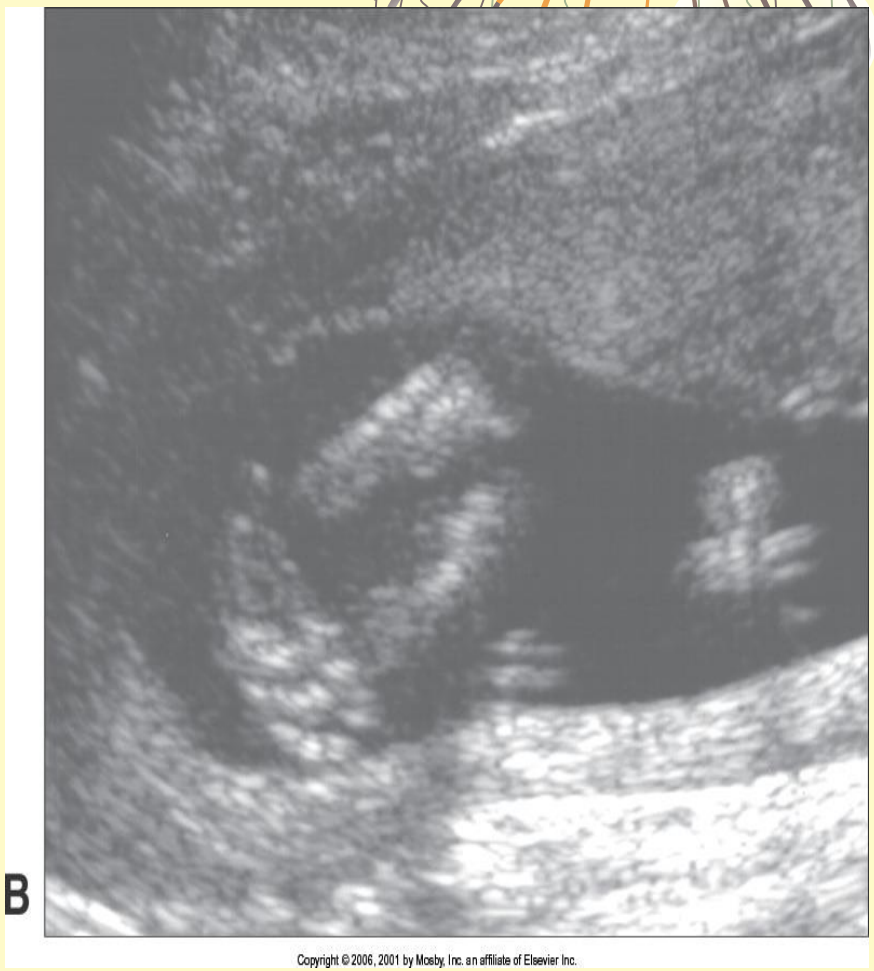
Foot Anomalies



- **Clubfoot (talipes)**
  - **Persistent abnormal inversion of the foot perpendicular to the lower leg**
- ***Rocker-bottom foot***
  - **Characterized by prominent heel and a convex sole**

Bilateral Talipes





Both feet inverted medially