Skeletal Dysplasia
Cased-Based Learning

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Radiologic Approach

- Assess Proportion
  - Rhizo-, meso-, or acromelia
  - +/- platyspondyly

- Assess Components of Bone
  - Epiphyses small or irregular ➔ epiphyseal dysplasia
  - Metaphyses widened, flared, or irregular ➔ metaphyseal dysplasia
  - Diaphyses widened or thickened ➔ diaphyseal dysplasia
Achondroplasia Group

- All have abnormalities of the same chromosomal locus and gene product, fibroblast growth factor receptor 3 (FGFR3)
  - Thanatophoric dysplasia
  - Achondroplasia
  - Hypochondroplasia
Thanatophoric Dysplasia

- AD
- Probably the most common lethal bone dysplasia
Thanatophoric Dysplasia
Thanatophoric Dysplasia

- Skull – *kleeblatschädel* in type II
Thanatophoric Dysplasia

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- Thorax - very short ribs and handlebar clavicles
Thanatophoric Dysplasia

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Thanatophoric Dysplasia

- Spine – small flat vertebral bodies with round anterior ends, U or H-shaped on AP
Thanatophoric Dysplasia
Thanatophoric Dysplasia

- **Pelvis**
  - Small, flared iliac bones
  - Very narrow sacrosciatic notches, flat dysplastic acetabula

- **Extremities – telephone receiver femora**
Thanatophoric Dysplasia
Thanatophoric Dysplasia
Case 1 – 5 yo with short stature
Achondroplasia

• Most common nonlethal skeletal dysplasia
• AD, spontaneous mutation rate 80%
Achondroplasia

• Skull
  – Large with midface hypoplasia
  – Small skull base and foramen magnum
Achondroplasia

- **Spine**
  - very short pedicles – risk of spinal canal stenosis
  - Decrease in interpediculate distance – lumbar spine
Achondroplasia

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  - very short pedicles – risk of spinal canal stenosis
  - Decrease in interpediculate distance – lumbar spine
Achondroplasia

- Pelvis
  - Elephant-ear iliac wings
  - Flat acetabular roofs
  - Narrow sacrosciatic notches
Achondroplasia

• Extremities
  – Rhizo- > meso- and acromelia
  – Hands – brachydactyly with metaphyseal cupping of MC’s
  – Knees – chevron and inverted chevron deformities
  – Hips proximal femoral fade out and hemispheric capital femoral epiphyses
Achondroplasia

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Case 2 – Newborn with severe respiratory distress
Short Rib-Polydactyly Group

• Includes
  – SRP I-IV - some with, some without polydactyly
  – asphyxiating thoracic dysplasia
  – chondroectodermal dysplasia

• Shortest ribs of all dysplasias
Short Rib-Polydactyly

- Thorax – shortest ribs, horizontal ribs
Short Rib-Polydactyly

- Thorax – shortest ribs, horizontal ribs
- Pelvis – small ilia, notched acetabula
Short Rib-Polydactyly

• Extremities
  – Micromelia
  – Rolling pin-shaped or round-ended or spiked femora
  – Ovoid tibiae
  – Polydactyly in some types
Asphyxiating Thoracic Dysplasia (Jeune Syndrome)

• Mixed prognosis
  – Some succumb early from respiratory compromise
  – Others die later from progressive nephropathy
Asphyxiating Thoracic Dysplasia (Jeune Syndrome)

- Thorax
  - Long and barrel-shaped
  - Handlebar clavicles
  - Short horizontal ribs with flared anterior ends
Asphyxiating Thoracic Dysplasia (Jeune Syndrome)

• Spine – normal
• Pelvis
  – Trident acetabular roof
  – Flared iliac wings
  – Narrowed SS notches
• Extremities – cone-shaped epiphyses in hands
Case 3 – 6 year old with history of congenital heart disease
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Chondroectodermal Dysplasia (Ellis-van Creveld Syndrome)

• Nonskeletal findings important in diagnosis
  – Hair, nail and teeth abnormalities
  – Congenital heart disease
Chondroectodermal Dysplasia (Ellis-van Creveld Syndrome)

- Thorax – small with short ribs
- Pelvis
  - Trident acetabula
  - Small, flared iliac wings
  - Narrowed SS notches
Chondroectodermal Dysplasia (Ellis-van Creveld Syndrome)

- Thorax – small with short ribs
- Pelvis
  - Trident acetabula
  - Small, flared iliac wings
  - Narrowed SS notches
Chondroectodermal Dysplasia (Ellis-van Creveld Syndrome)

• Extremities
  – Generalized shortening
  – Exostosis of proximal medial tibia
  – Post-axial polydactyly
  – Capitate-hamate fusion
  – Extra carpal bone
  – Cone-shaped epiphyses
Chondroectodermal Dysplasia (Ellis-van Creveld Syndrome)

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Case 4 – newborn boy with respiratory distress
Chondrodysplasia Punctata Group

- All have epiphyseal stippling
- Rhizomelic
  - AR, death in first year
  - Spine – coronal clefts
  - Symmetric bilateral shortening of femora
Chondrodysplasia Punctata Group

- Conradi-Hunermann
  - X-linked dominant
  - Asymmetric shortening of limbs
  - Diffuse stippling of the spine
Chondrodysplasia Punctata Group
Chondrodysplasia Punctata Group
Case 5 – 12 yo boy with short stature and unusual hair
Metaphyseal Chondrodysplasia Group

- All have normal spine and wide irregular metaphyses
- Jansen-type
  - Most severe
  - Infantile presentation
  - AD
  - Extremities – extensive irregular, expanded metaphyses
  - Hyperparathyroidism
Metaphyseal Chondrodysplasias

- Schmid-type – mildest, metaphyseal flaring, especially around knees
- Schwachman-Diamond – AR
  - Pancreatic insufficiency – malabsorption and lipomatosis of pancreas
  - Cyclic neutropenia – recurrent infections
Metaphyseal Chondrodysplasias

- McKusick-type
  - Cartilage-hair hypoplasia
  - High frequency in the Amish and Finnish populations
Metaphyseal Chondrodysplasias

• McKusick-type
  – Hirschsprung disease
  – Immune deficiency and increased risk of malignancy, especially leukemia and lymphoma
Metaphyseal Chondrodysplasias

• McKusick-type
  - Spine – square vertebral bodies
  - Extremities – flaring, cupping and fragmentation of metaphyses, especially at the knees
  - Hands – shortening with metacarpal and phalangeal cupping and coning
Metaphyseal Chondrodysplasias

[Images of X-rays showing typical metaphyseal changes]
Metaphyseal Chondrodysplasias

- McKusick-type
Case 6 – Fretful 8 mo whose pediatrician thinks he has bilateral clavicular fractures
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Dysplasias with Prominent Membranous Bone Involvement

• Cleidocranial dysplasia
  – AD, marked variability in expression
  – Drooping narrow chest, hypermobile shoulders, and dental anomalies
  – Mild short stature
Cleidocranial Dysplasia

- Skull – wormian bones and wide, open anterior fontanelle
Cleidocranial Dysplasia

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Cleidocranial Dysplasia

- Thorax – hypoplasia or absence of clavicles, downward sloping ribs
Cleidocranial Dysplasia

- Spine – posterior wedging of vertebral bodies
Cleidocranial Dysplasia

- Pelvis - high, narrow iliac wings, absence or hypoplasia of pubic bones
Case 7 – short 3 yo with unusual facial appearance
Dysostosis Multiplex Group

- Mucopolysaccharidoses and mucolipidoses
- All AR
- All produce similar radiographic complex of findings
Hurler Syndrome

• Present in infancy or early childhood
• Skull – J-shaped sella
Hurler Syndrome

• Thorax
  – Short thick clavicles
  – Oar-shaped ribs
Hurler Syndrome

• Spine
  – Gibbus deformity
  – Inferior beaked T-L vertebral bodies
Hurler Syndrome

- Pelvis – small flared iliac wings with inferior tapering and steep acetabular roofs
Hurler Syndrome

• Extremities
  – Wide diaphyses of long bones and metacarpals
  – Pointed proximal metacarpal poles
Hurler Syndrome
Morquio Syndrome

- Dens hypoplasia/atlantoaxial instability
- No J-shaped sella
- Vertebral beak is in the middle
- Ribs are widened but not oar-shaped
- Proximal metacarpal poles are rounded
Case 8 – 31-week fetus
Dysplasias with Decreased Density

• Very large group of conditions that share an abnormality of type I collagen
Osteogenesis Imperfecta Type II

- Invariably lethal
- Skull – poor or absent ossification
- Thorax – small chest with beaded ribs
Osteogenesis Imperfecta Type II

- Invariably lethal
- Skull – poor or absent ossification
- Thorax – small chest with beaded ribs
Osteogenesis Imperfecta Type II

- Spine – very poor ossification with collapse of vertebral bodies
- Extremities – accordion femora
Lethal Bone Dysplasias

- Thanatophoric dysplasia
- Asphyxiating thoracic dysplasia (usually)
- Osteogenesis imperfecta, type II
Case 9 – 17 year old with short stature
Pyknodysostosis

- AR, presents in infancy
- Micrognathia, short fingertips, fractures
- Generalized osteosclerosis
Pyknodysostosis

- Skull
  - Wormian bones
  - Marked delay in closure of sutures and fontanelles
Pyknodysostosis

- Skull
  - Obtuse mandibular angle
Pyknodysostosis

- Skull
  - Obtuse mandibular angle
Pyknodysostosis

- Thorax – resorption of acromial ends of clavicles
Pyknodysostosis

- Spine - spondylolisthesis
Pyknodysostosis

- Extremities – resorption of phalangeal tufts
- Extremities - fractures
Wormian Bones

- **Common**
  - Normal – up to 8-10 small
  - OI
  - Cleidocranial dysplasia
  - Hypothyroidism

- **Uncommon**
  - Pycnodysostosis
  - Menke’s kinky hair
  - Hypophosphatasia