Skeletal Dysplasias

TOP TEN THINGS TO KNOW

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Skeletal Survey

- Diagnostic tool for evaluating skeletal dysplasias and syndromes

- Narrow the DDx, guiding genetic testing and treatment and prognosis
• AP and Lateral Skull
• Lateral Cervical Spine
• AP and Lateral Thoracolumbar spine
• AP chest
• AP Pelvis
• AP all extremities (to include hands and feet)

• **Pearl:** Get true lateral view of c-spine (fusion, vertebral shape, odontoid)

**Skeletal Survey**
- Sutures
- Wormian Bones
- Sella Turcica
- Craniofacial Proportions
- Calvarial/Skull base thickening

Skull
J-shaped sella
J-shaped sella
• Alignment
• Vertebral Shape
• Fusion/Segmentation Anomalies
• Interpediculate narrowing lower lumbar spine

Spine
Nomenclature for abnormal vertebral body shape - lateral view

• Normal vertebra
• Posterior Scalloping
• Endplate irregularity

• Platyspondyly
• Anterior Beaking
• or
Nomenclature for abnormal vertebral body shape - lateral view

- Normal vertebra
- Short Pedicles
- Coronal Clefting
- Anterior Wedging
- Posterior Wedging
- Tall Vertebral Body
Narrowing of Interpediculate Distance

- Normal vertebral column - AP

- Decreased interpediculate distance

Moving inferiorly in the lumbar spine, pedicles become closer together
• Sciatic Notches
• Acetabular Roofs
• Iliac Wings
• Pubic bone ossification

Pelvis
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Pelvis

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Pelvis
• Focal or Generalized?
• Epiphysis, Metaphysis, Diaphysis?
• Small epiphyseal ossification centers
• Metaphyseal flaring, marginal irregularity

Extremities
Extremities

- Disproportionate shortening
  - Rhizomelia (proximal)
  - Mesomelia (middle)
  - Acromelia (distal)
- Micromelia
- Polydactyly, duplications, hypoplasia
Rhizomelia

- Disproportionate shortening of proximal aspect of long bones (humerus, femur)
- Achondroplasia
• Most common non-lethal skeletal dysplasia
• FGFR3 mutation (also thanatophoric dysplasia, hypochondroplasia)
• Abnormal enchondral bone formation
• **Skull**- Frontal bossing, midface hypoplasia
• **Spine**- Interpediculate narrowing, posterior scalloping, short pedicles
• **Thorax**- Mildly short ribs
• **Pelvis**- Flat acetabular roofs, small sciatic notches, short and wide iliac wings
• **Extremities**- Rhizomelia

Achondroplasia
Achondroplasia
- Thanatophoric Dysplasia
- Jeune Syndrome (Asphyxiating Thoracic Dysplasia)

Lethal Dysplasias
Similarities with Achondroplasia:
- FGFR3 mutations
- Pelvis
- Narrow foramen magnum
- Interpediculate narrowing L spine
Thanatophoric Dysplasia
• Differences with Achondroplasia:
  • Shorter ribs
  • Micromelia (all portions short) with bent bones
  • More platyspondyly
  • LETHAL

Thanatophoric Dysplasia
Jeune Syndrome
Jeune Syndrome
• Asphyxiating Thoracic Dysplasia (a.k.a.)

• Normal Spine!
• Irregular Acetabular Roof (Trident shaped)
• No bent bones!
• Short ribs! (variably lethal)

Jeune Syndrome
• “Dysostosis Multiplex”
• J-shaped sella (anterior flattening)
• Anterior beaking vertebra T-L junction
• Proximal pointing metacarpals
• Thickened ribs
• Narrow inferior portion of iliac bones

Mucopolysaccharidoses
• Resources:
  • Textbooks (Taybi and Lachman)
  • Google groups
  • Interactive Atlas - Seattle Children’s radiology website (free)
  • ibook - Little Book of Skeletal Dysplasias (free)
• Multidisciplinary conferences (ortho, genetics, endocrine, rads)
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You are not alone!