Imaging of Generalized Skeletal Disorders

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Importance

- Common disorders
  - 1 in 3,000 to 5,000 births (450 dysplasias)
- Familial
- Lifelong diseases
- New and exciting developments
- New therapies
Spondylocostal Dysostosis

- Skeletal findings signal more important problems
- Chest deformities: respiratory failure
Importance: Duration of Disease

• Lifelong diseases
  – Prognosis
  – Detection of complications
  – Quality of life
  – Genetic counseling
  – Screening
Importance: New and Exciting Developments

• Mapping of genetic defects (>300/450)
  – New phenotypes
• Newer imaging technologies
  – Insight into cartilaginous and soft tissue abnormalities
Importance: New Therapies

- Limb lengthening
- Bone marrow transplantation
  - Mucopolysaccharidosis
  - Osteopetrosis
- Gene therapy
Importance: Radiology’s Role

• Radiology remains crucial for diagnosis and management
• Declining skills in our subspecialty!
Current Diagnosis

• Physical examination
• Radiographs (Genetic Skeletal Survey)
  – AP & lateral views of skull, entire spine
  – AP of pelvis, 4 extremities, hands & feet
• Genetic studies
• Biochemical studies
  – Mucopolysaccharides
Generalized Skeletal Disorders
Nosology and Classification of Genetic Skeletal Disorders (2010)

- Osteochondrodysplasias
- Dysostoses (individual bones)
- Chromosomal abnormalities
- Primary metabolic abnormalities
- Tumoral disorders
- Osteolysis
Some Osteochondrodysplasia Families

1. FGFR3 (Achondroplasia)
2. Type 2 Collagen (SED)
3. Sulphation disorders (Diastrophic dysplasia)
4. Short rib dysplasia (Jeune)
5. Multiple epiphyseal dysplasia group (MED)
6. Metaphyseal dysplasia group (Schmid)
Heterozygous Achondroplasia

- Large calvarium, frontal bossing
- Small skull and foramen magnum
Heterozygous Achondroplasia

• Large calvarium, frontal bossing
• Small skull and foramen magnum
• Small spinal canal
Heterozygous Achondroplasia

- Large calvarium, frontal bossing
- Small skull and foramen magnum
- Small spinal canal
- Square iliac bones
- Narrow sacrosciatic notches
Heterozygous Achondroplasia

- Lucent proximal femurs
- Flared metaphyses with central cupping
- Trident hand

* Decreased cartilage proliferation with near normal morphology
Osteochondrodysplasia Families

- Achondroplasia (FGFR3)
- Diastrophic dysplasia
- Collagenopathies
Neurofibromatosis

• Most common single gene disease
• Neurofibromas
  – Plexiform along nerves
  – Dermal
  – Characteristic MR appearance
• Dysplasia
  – Thin, ribbon-like bones
Prenatal Diagnosis

• Radiography
• Sonography
  – Increased brain visualization (OI, hypophosphatasia)
  – Bowed bones (thanatophoric)
  – Fractures (OI)
• MRI- echo planar imaging
• Low dose CT
Thanatophoric Dysplasia
Osteogenesis Imperfecta (Sillence)

I- Most common, few fractures at birth, blue sclera
II- Greatest # of fractures, usually broad bones (accordion)
III- Multiple fractures, bowed bones
IV- White sclera, few fractures, very rare
Type I
Wormian Bones

- Normal
- OI
- Cleidocranial dyostosis
- Progeria
Cleido-cranial dysplasia
Type II
Type III
Popcorn Calcifications
OI: Post-therapeutic changes
Involved Segment

- Epiphyseal
- Metaphyseal
- Diaphyseal
Epiphyseal Involvement

• Misshapen before birth: Trevor

• Irregular in first years of life:
  – Spondyloepiphyseal dysplasia
  – Diastrophic dysplasia
Epiphyseal Involvement

- Delayed ossification: SED
Stippled Epiphysis

- Associated with abnormal growth
- Chondrodysplasia punctata
- Other dysplasias
- Warfarin
Physeal Involvement

• Slowdown: achondroplasia
• Widening
  – No ZPC: rickets
  – Preserved ZPC: Schmid
Loss of ZPC-
Hypophosphatematic rickets
Hypophosphatemia
Metaphyseal Involvement
Spine Involvement in Dysplasia

- Decreased interpediculate distances:
  - Decreased endochondral growth
  - Associated with narrow pelvis
Spine Involvement

- Irregular endplates
  - Spondyloepiphyseal dysplasia, Mucopolysaccharidosis
- Beaking
  - Mucopolysaccharidosis
Clefts – Cleidocranial Deysplasia
C1-2 Instability

- Trisomy 21
- Muco-polysaccharidosis
- SED
Segmental Classification of Dysplasias

- Rhizomelic (proximal)
  - achondroplasia
- Mesomelic (middle)
  - Dyschondrostosis: Madelung
- Acromelic (distal)
  - Chondroectodermal dysplasia
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Asphyxiating Thoracic Dystrophy (Jeune)

- Family of polydactyly with thoracic restriction
Generalized Skeletal Disorders

• Continue to be a challenge
• RELEVANT