SKELETAL DYSPLASIA

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INITIAL EVALUATION

History and Physical Exam

Developmental motor delay, loss of function, neurological complaints
Family history

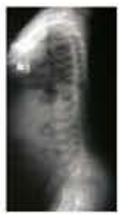
Facial features; spinal deformity; limbs-joint laxity, deformity; neurological exam; proportion- short vs. average trunk length; limb shortening- rhizo, meso or acromelia

Typical Radiographs

Flexion and extension lateral C-spine, AP and lateral thoracolumbar spline, AP and lateral lower extremities hip to ankles, AP both hands and wrists

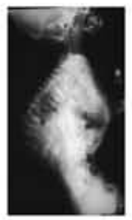
4 yo fraternal twins, one with achondroplasia. Note rhizometic limb shortening and normal trunk length

Short trunk in Morquio's syndrome



Spondyleopiphyseal Dysplasia

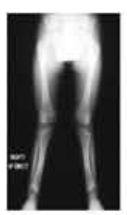
Platyspondyly
Morquio's Syndrome



Metatropic Dysplasia

Areas of Limb Involvement

Spondyleopiphyseal dysplasia. Primarily epiphyseal involvement.



Chondrectodermal dysplasia. Selective tibial epiphyseal involvement.

Metaphyseal dysplasia. Note similarity to rickets.

Metatropic Dysplasia. Epiphysis and metaphysis involved.

ACHONDROPLASIA

Etiology

FGFR3 (fibroblast growth factor receptor 3)
Autosomal Dominant

Diagnosis

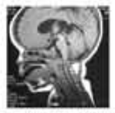
Clinical diagnosis at birth can be confirmed using lab tests and radiological features

Clinical Features

Average length and weight at birth
Macrocephaly, Mid-face hypoplasia
Rhizomelia
Genu varum
Trident hand

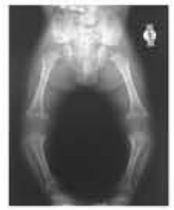
Clinical Problems

Foramen magnum stenosis
Sleep apnea
Hydrocephalus



Thoracolumbar kyphosis usually resolves
Frequent otitis media
Genu varum
Lumbar and cervical spinal stenosis
Obesity
Rarely needs joint replacement

Narrowing interpediculate distance
Champagne glass pelvis



Rhizomelia, Notched (V shaped) physes

SPONDYLOEPIPHYSEAL DYSPLASIA

Etiology

Multiple subtypes:
most common – congenital
Type II collagenopathy

Diagnosis

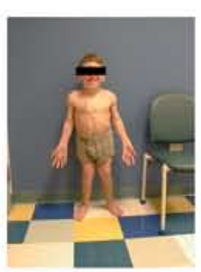
At birth; Short trunk, short limb
Radiographic features

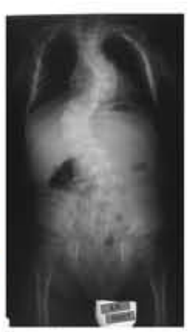
Clinical Features

Extreme short stature
Long, slim fingers
Waddling gait
Lumbar lordosis
Genu valgum

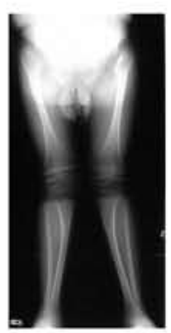
Clinical Problems

Cervical instability
Kyphoscoliosis
Coxa vara
Myopia, retinal detachment
Eventual joint replacement





Platyspondyly, Scoliosis



Severe epiphyseal dysplasia, coxa vara

MULTIPLE EPIPHYSEAL DYSPLASIA

Etiology

COMP (collagen oligomeric matrix protein) abnormality
Type IX collagen (less severe)
Autosomal dominant

Diagnosis

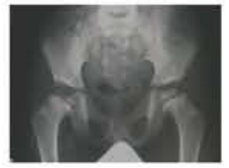
Radiographic features
Not usually recognized until age 5-10 yrs

Clinical Features

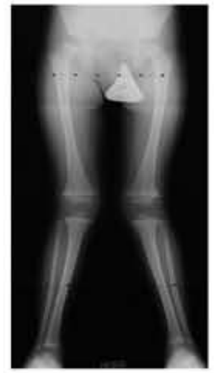
Mild short-stature
Delayed walking
Genu valgum

Clinical Problems

Painful, stiff joints
Limb malalignment
Eventual joint replacement



Appearance of bilateral Legg-Perthes is common in MED



Delayed epiphyseal ossification and small epiphyses

DIASTROPHIC DYSPLASIA

Etiology

Sulphate transporter defect
Autosomal recessive

Diagnosis

At birth; cleft palate, clubfeet, hitch hiker thumb,
Symphalangism, cauliflower ears develop early

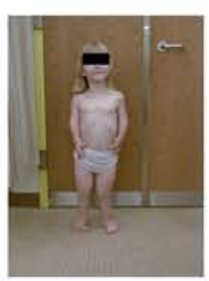
Clinical Features

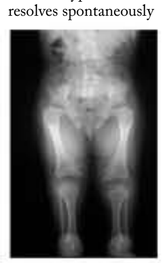
Short stature
Flexion contractures – hips knees
Lateral patellar dislocation
Foot deformity

Clinical Problems

Laryngotracheomalacia
Cervical kyphosis
Kyphoscoliosis
Hip dysplasia
Limb Malalignment
Joint replacement common

Cervical kyphosis often resolves spontaneously





Short, thick tubular bones, delayed and fragmented epiphyses, hip dysplasia

PSEUDOACHONDROPLASIA

Etiology

COMP (collagen oligomeric matrix protein) abnormality
Autosomal dominant

Diagnosis

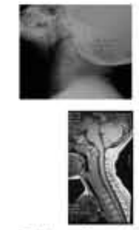
Radiographic features
Short-limbed dwarfism not usually recognized until 2-3 yrs

Clinical Features

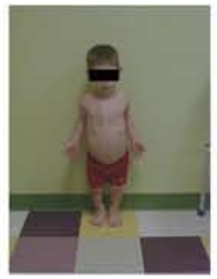
Normal face
Short, stubby, hyper mobile fingers
Genu varus/valus or windswept
Marked ligamentous laxity

Clinical Problems

Cervical spine instability
Limb malalignment
Eventual joint replacement



Radiographs and MRI – C1-2 instability with cord compression



Short broad long bones with flared metaphyses and small epiphyses

Platyspondyly

MORQUIO SYNDROME

Etiology

Mucopolysaccharidosis Type IVA
N-acetylgalactosamine-6-sulphatase deficiency
Autosomal recessive

Diagnosis

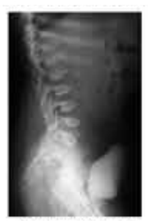
Not made at birth, short stature apparent by 2 yrs
Radiographic features
Urine screen — keratan sulfate
Molecular genetic testing

Clinical Features

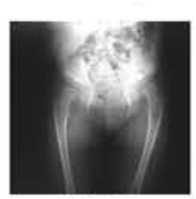
Normal intelligence
Awkward gait, genu valgum
Barrel-shaped chest, pectus carinatum
Extreme ligamentous laxity

Clinical Problems

Cervical spine instability (odontoid hypoplasia)
Limb malalignment
Corneal opacity (at 5-10 yrs)
Cardiomyopathy
Joint replacement common



Platyspondyly with central beaking



Hip dysplasia

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