SKELETAL DYSPLASIA

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INITIAL EVALUATION

History and Physical Exam
Developmental 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- rhiz, meso or acromelia

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

ACHONDROPLASIA

Etiology
FGFR3 (fibroblast growth factor receptor 3)
Autosomal Dominant

Clinical Features
Reduced head circumference
Mental retardation

Clinical Problems
Curvature of spine
Kyphosis
Flexion contracture of hips and knees

MULTIPLE EPIPHYSEAL DYSPLASIA

Etiology
Type III collagenopathy

Clinical Problems
Curvature of spine
Kyphosis
Flexion contracture of hips and knees

MORQUIO SYNDROME

Etiology
Mucopolysaccharidosis Type VII

Clinical Problems
CNS abnormalities
Cardiac abnormalities
Hepatic, renal, pulmonary

DIAGNOSIS

Typical Radiographs

Typically requires clinical recognition

Typical Radiographs

Typically requires clinical recognition

Typical Radiographs

Typically requires clinical recognition