Bone Dysplasia

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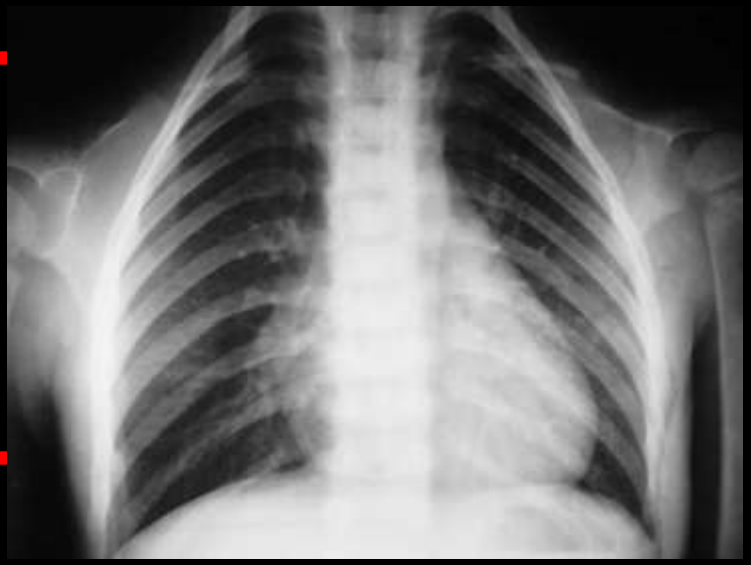


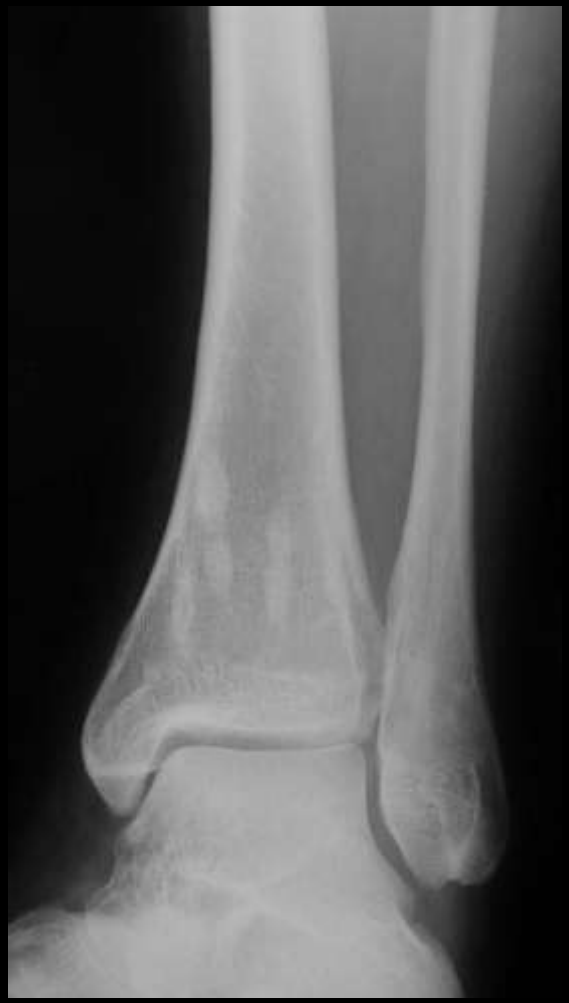
Benign Sclerosing Bone Dysplasia

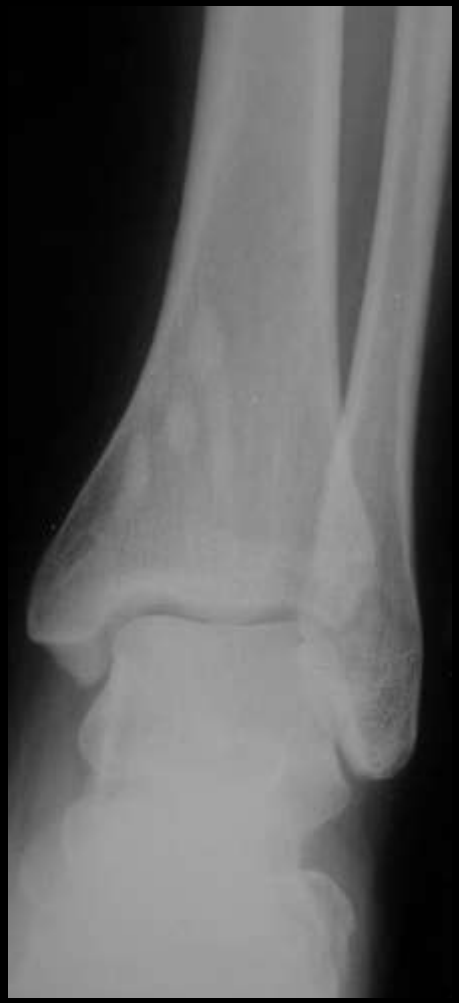
- Osteopoikilosis
- Melorrheostosis
- Osteopathia Striata

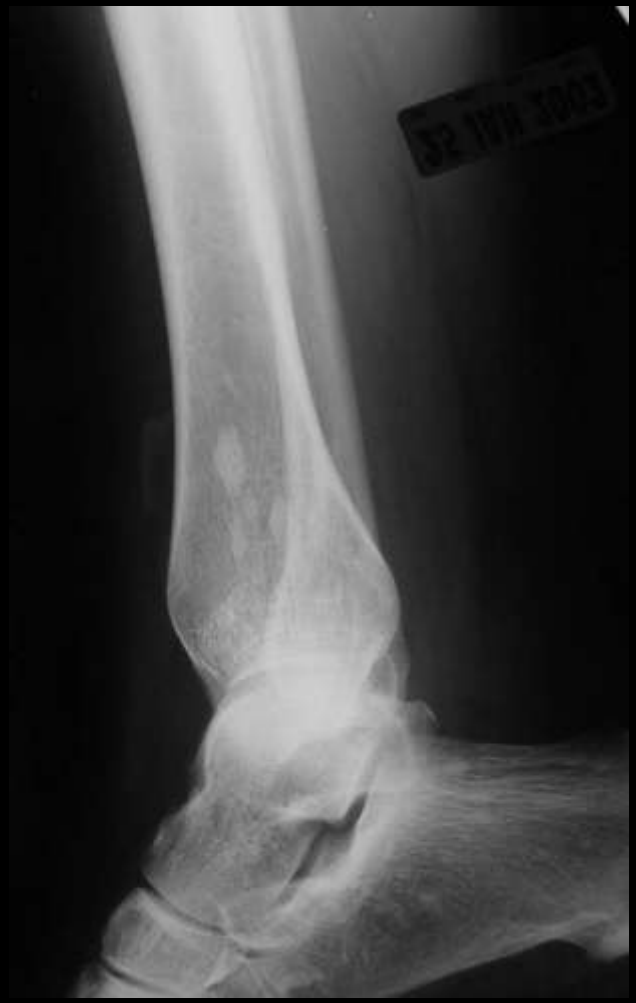
Osteopoikilosis

- Multiple bone islands
- Epiphyses > Metaphyses
- 1-10mm
- Appendicular and Pelvis
- Parallel to long axis of bone
- Skull, Spine and ribs spared

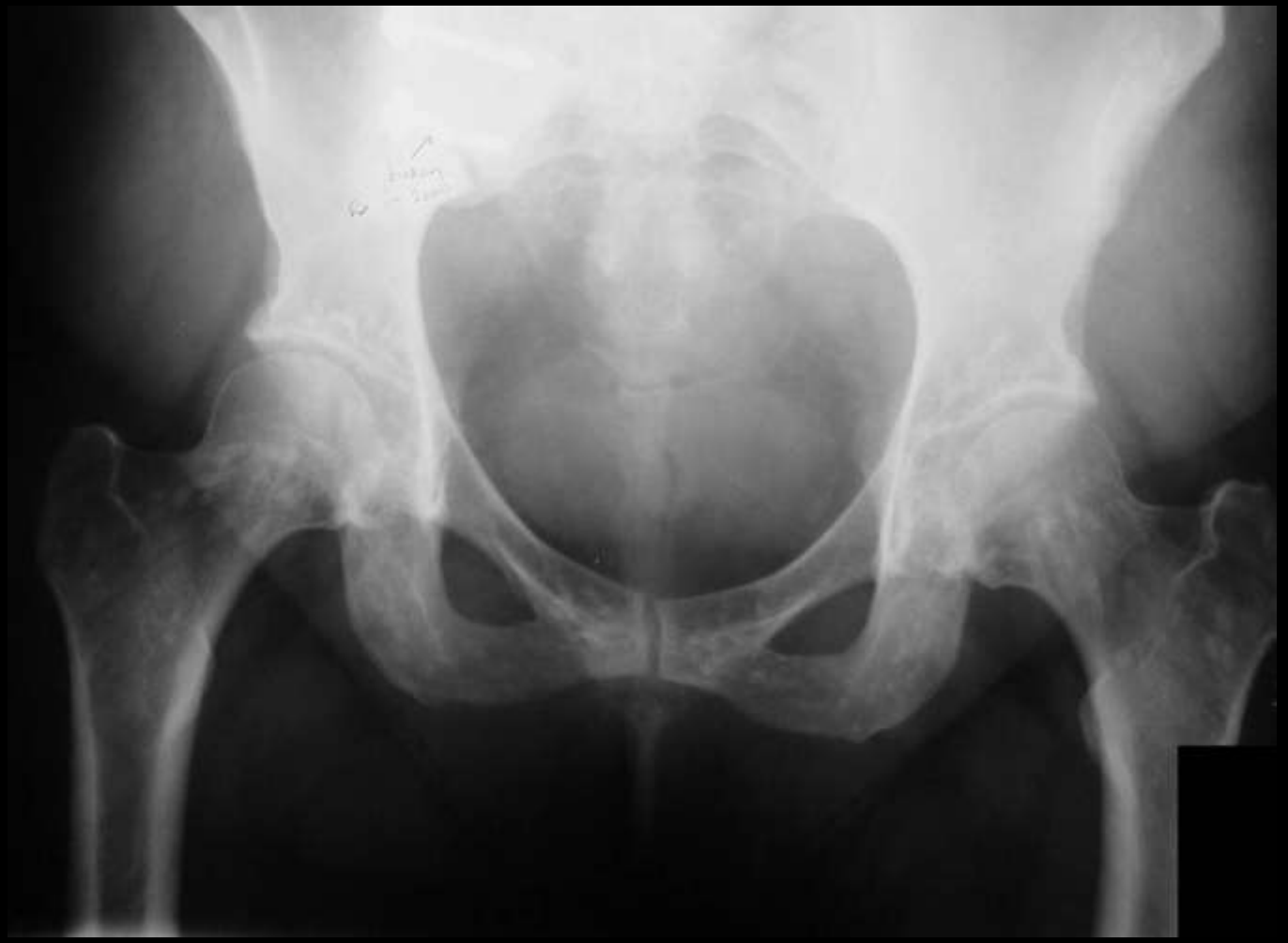
Osteopoikilosis







Osteopoikilosis



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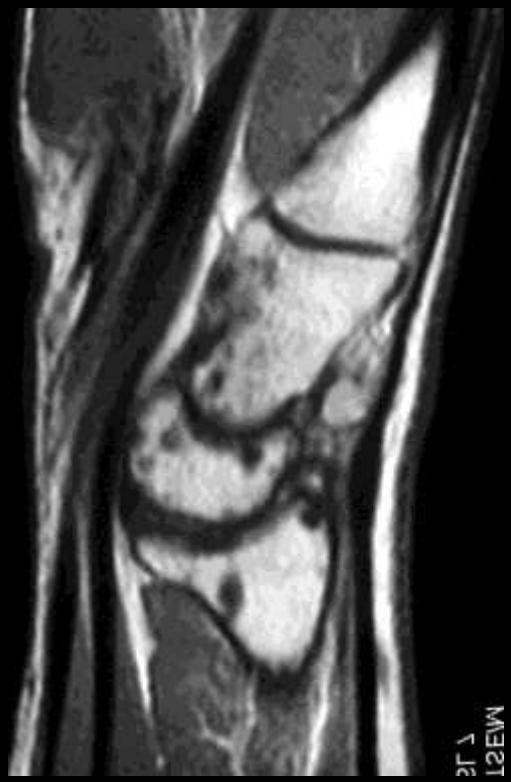
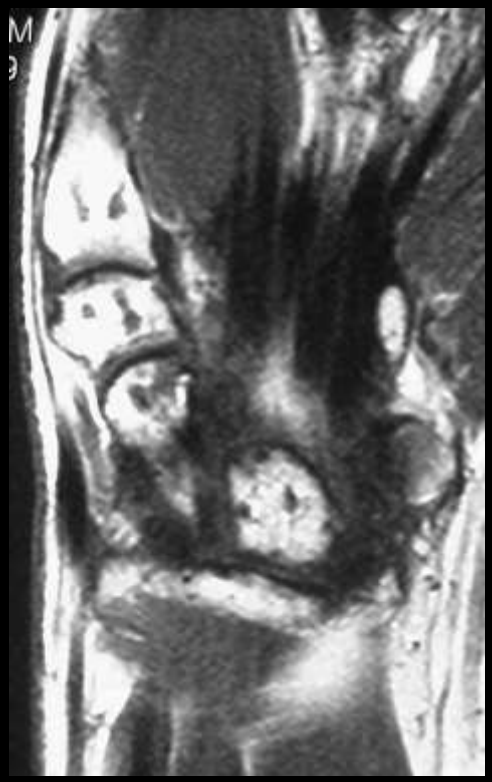
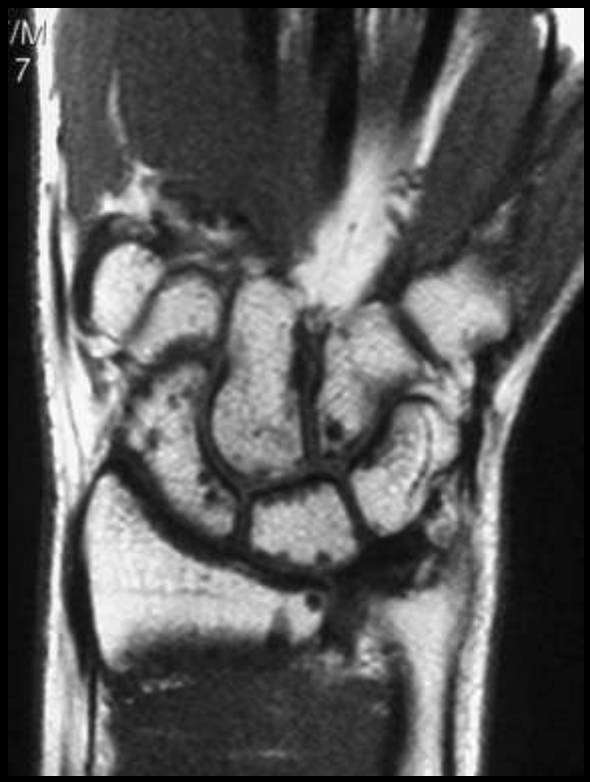


Saq T1





Cor T1

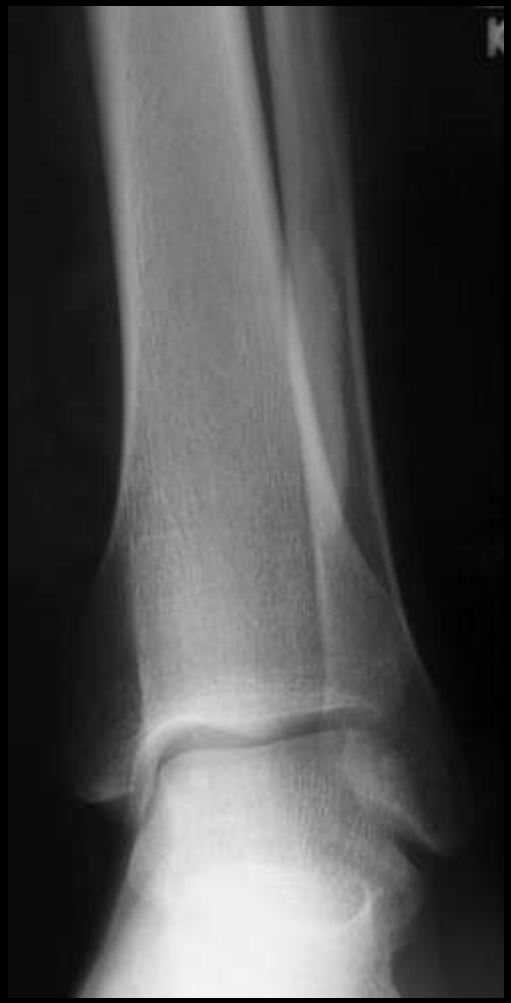
Multiple Sclerotic Bone Lesions

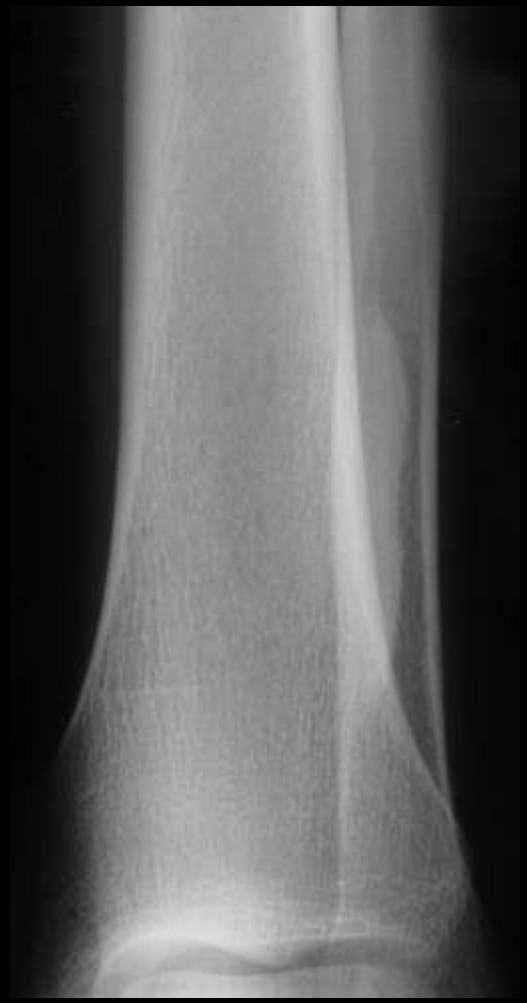
- Developmental
  - Bone islands
  - Fibrous dysplasia
  - Osteopoikilosis
  - Osteopathia Striata
  - Tuberous Sclerosis
- Neoplastic
  - Metastases
  - Lymphoma
  - Mastocytosis
  - Healing benign or malignant lesions
  - Myeloma
  - Osteomata
  - Multifocal osteosarcoma
- Idiopathic
- Vascular
- Traumatic

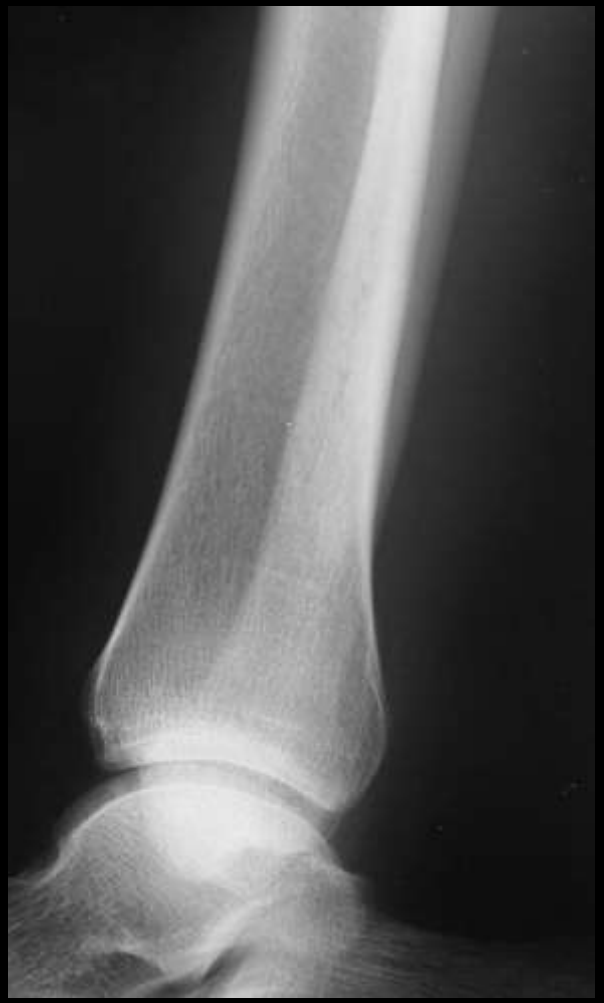
Melorheostosis

- Molten candle wax
- Cortical and Periosteal
- Extend from bone to bone
- Usually one limb
- Occ bilateral and asymmetric
- Skull spine and ribs seldom
- Sclerotome
- Occ ST
- Spinal associated with lipoma and myelocele

Melorrheostosis

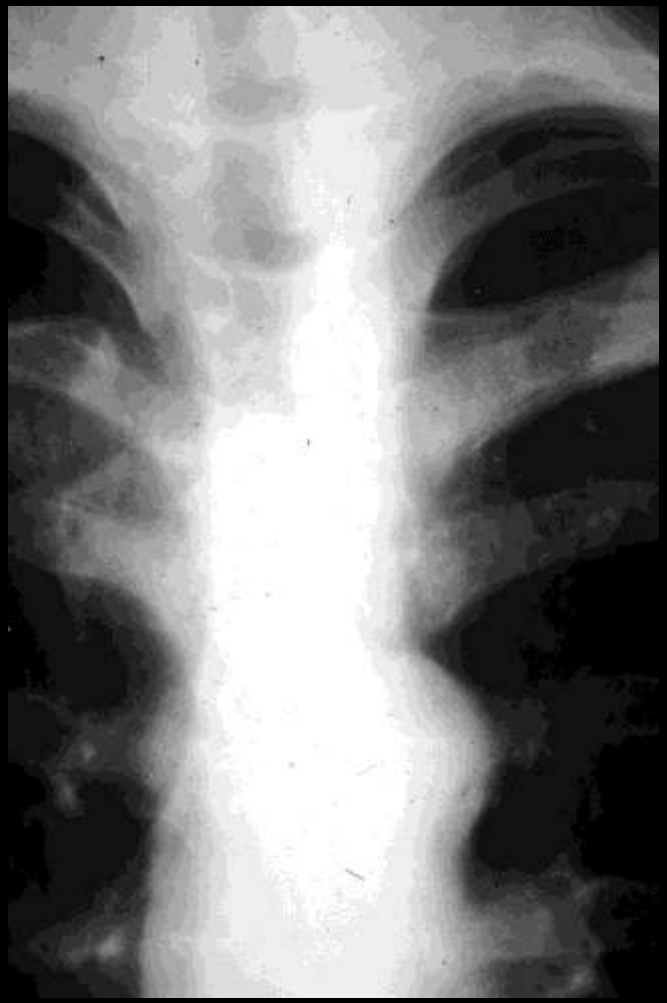


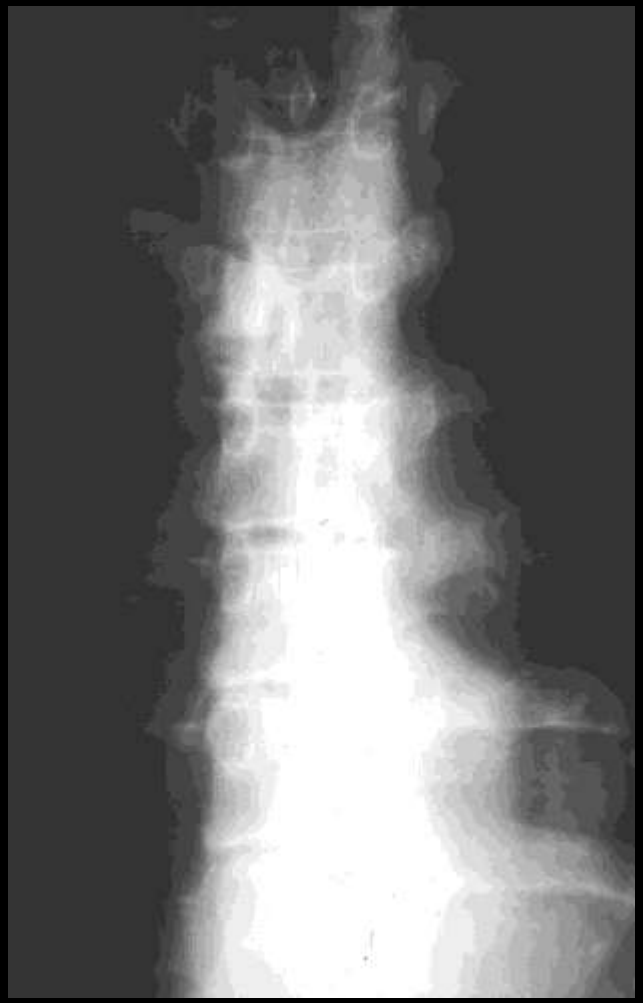




25M

Melorheostosis





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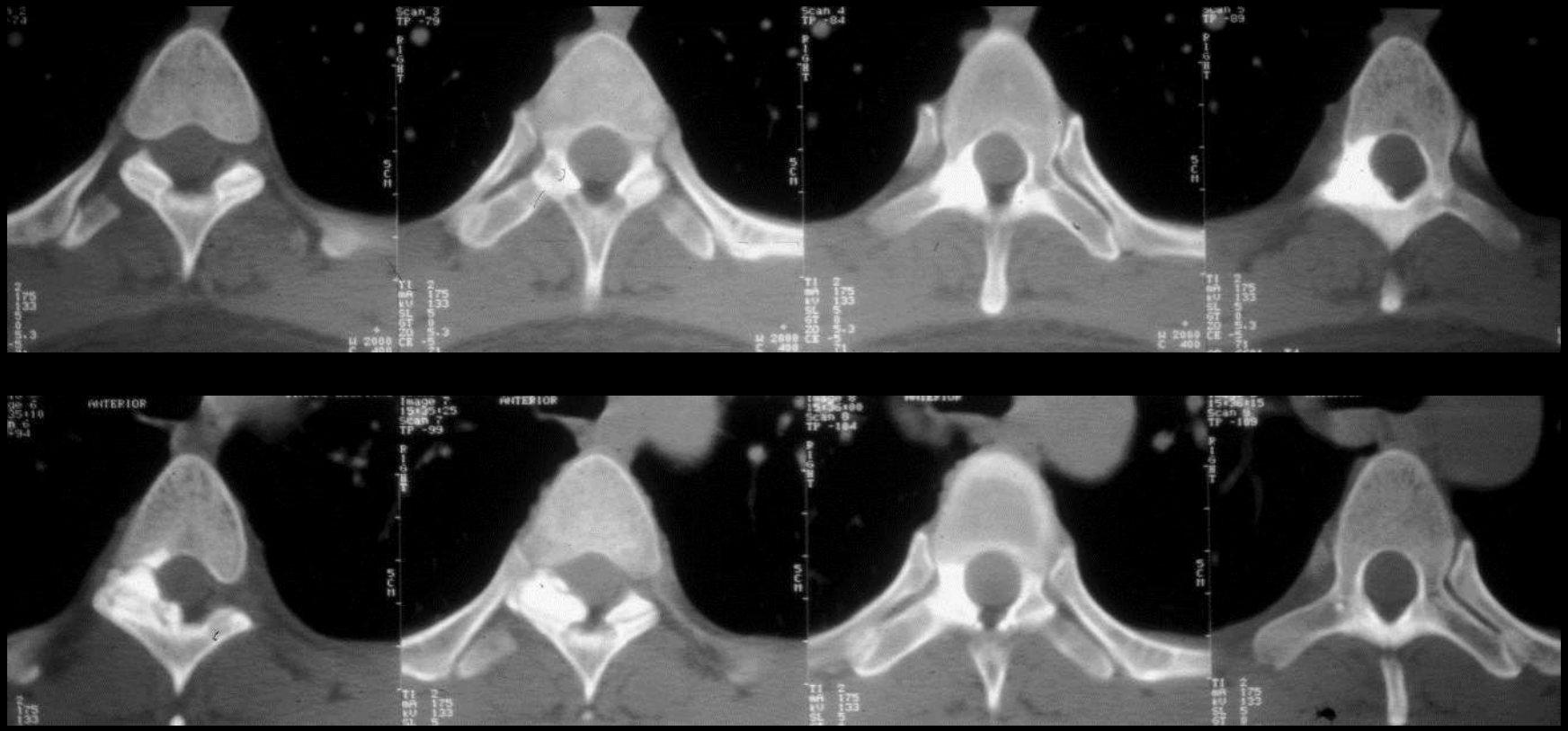

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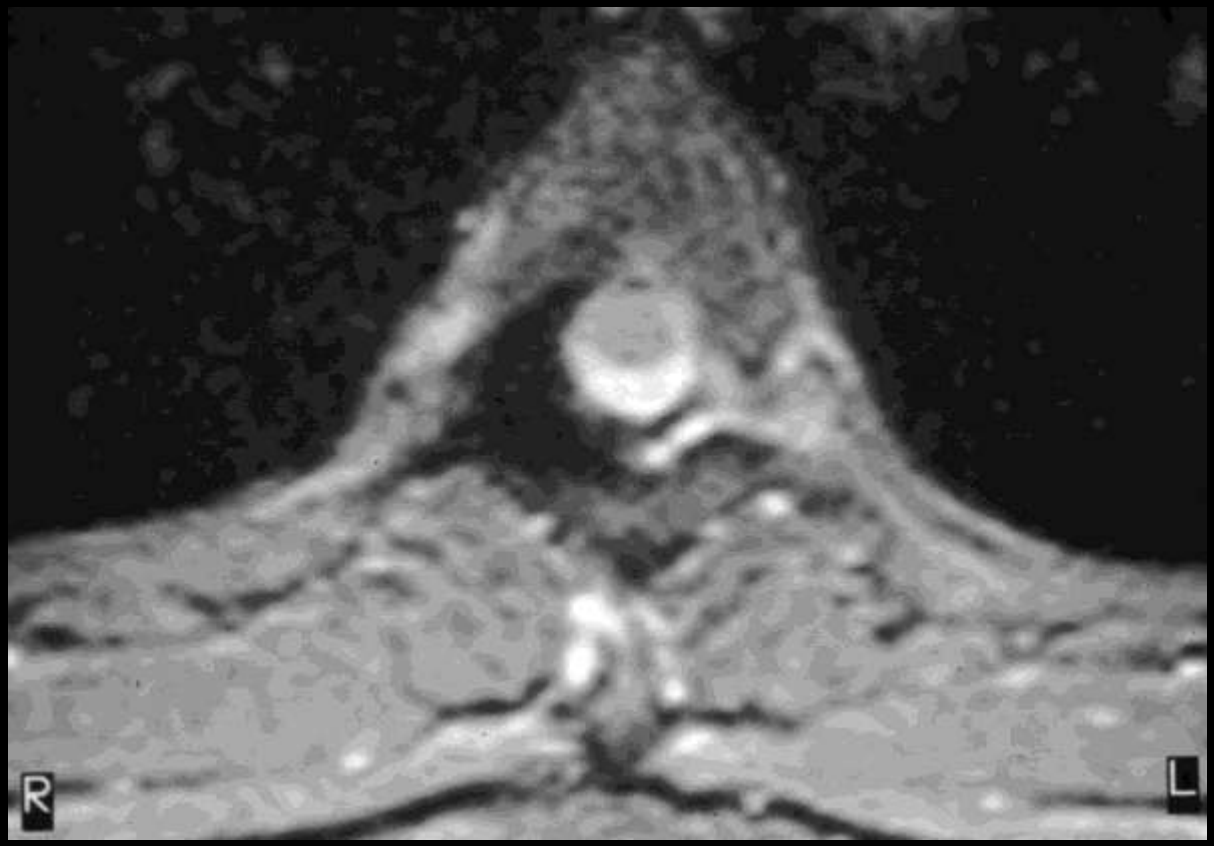

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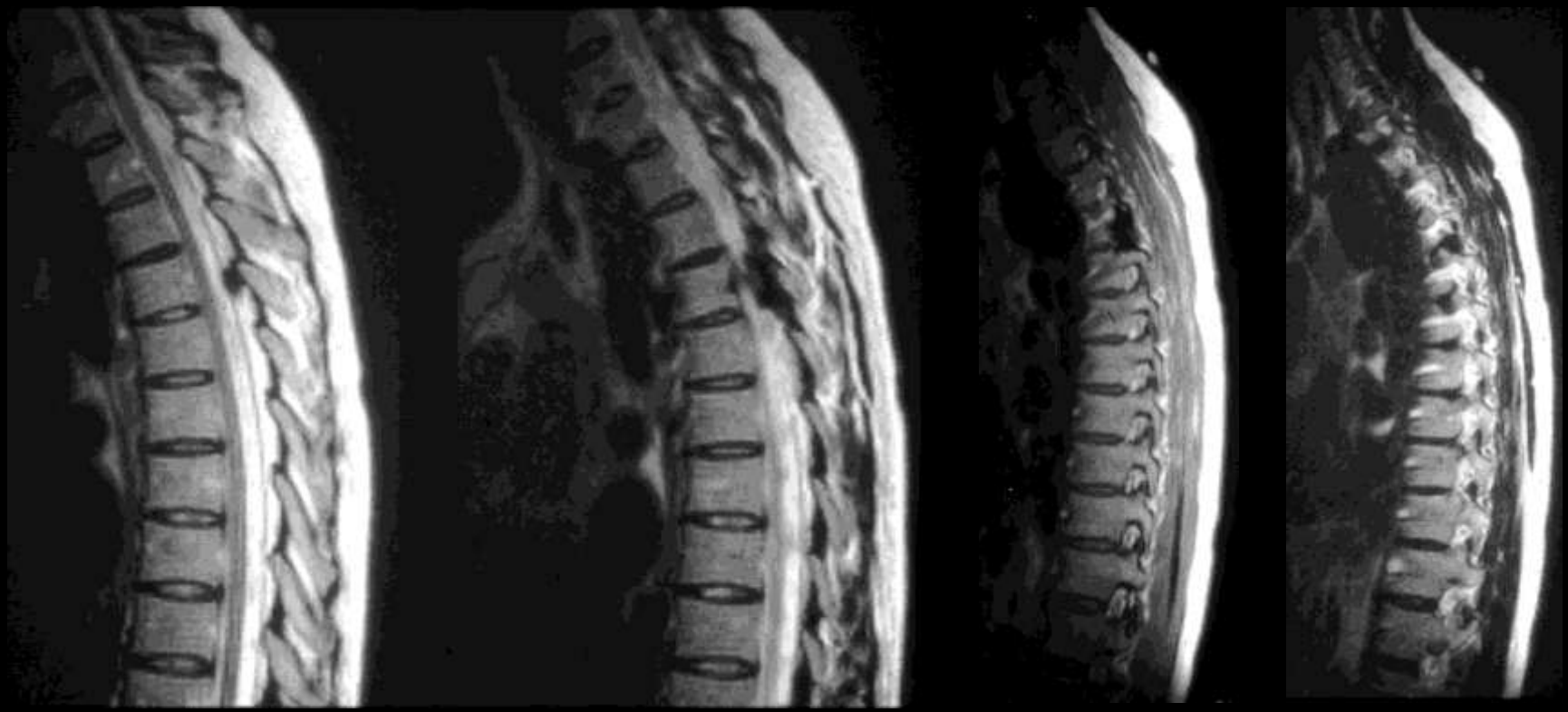

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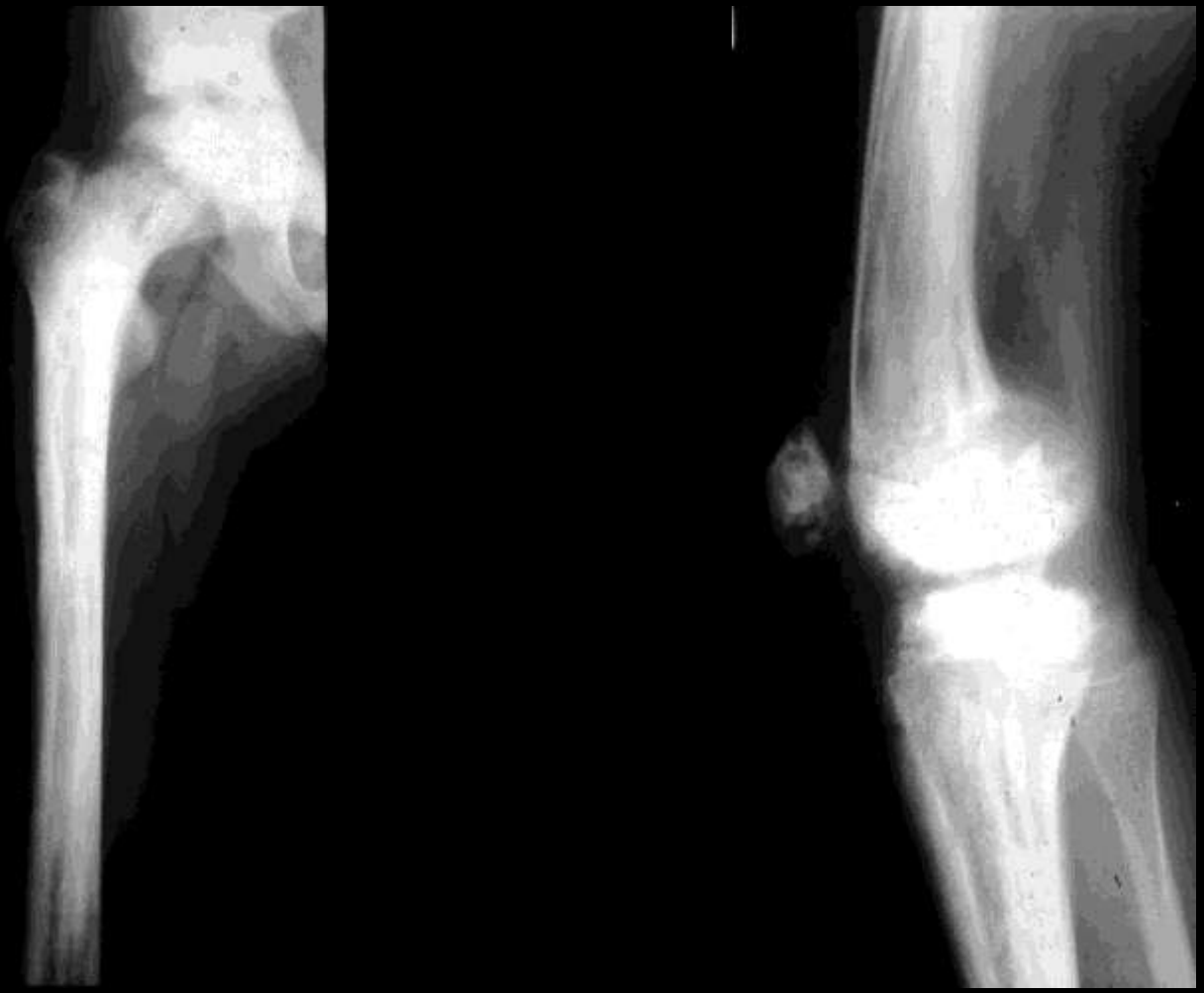
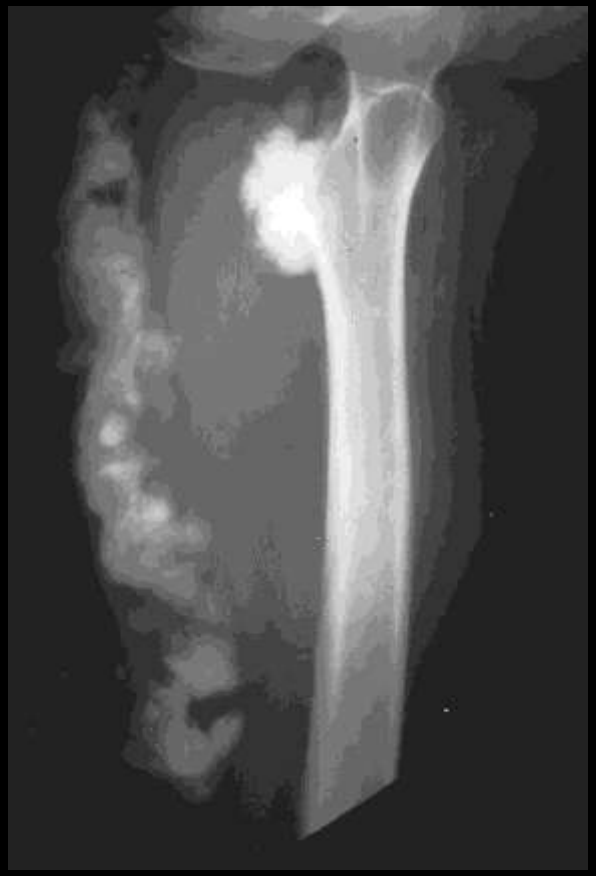
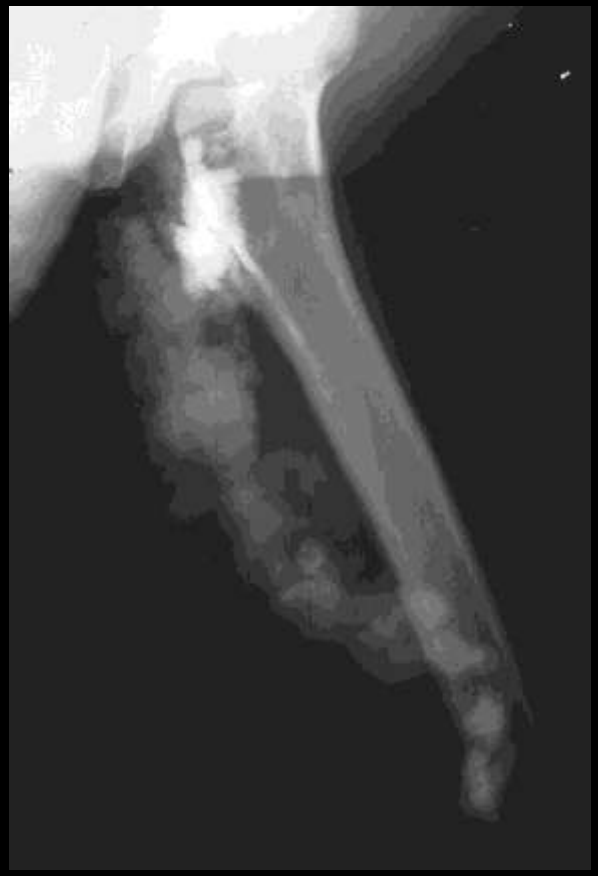
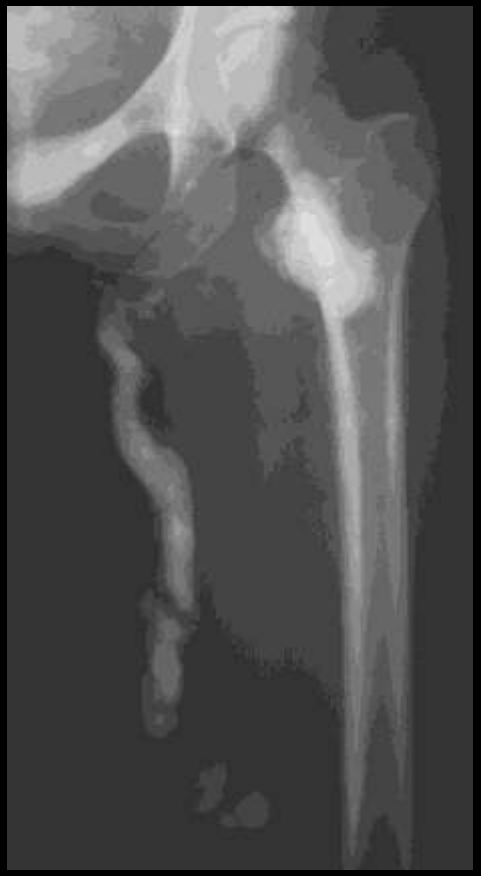


13M

Melorheostosis







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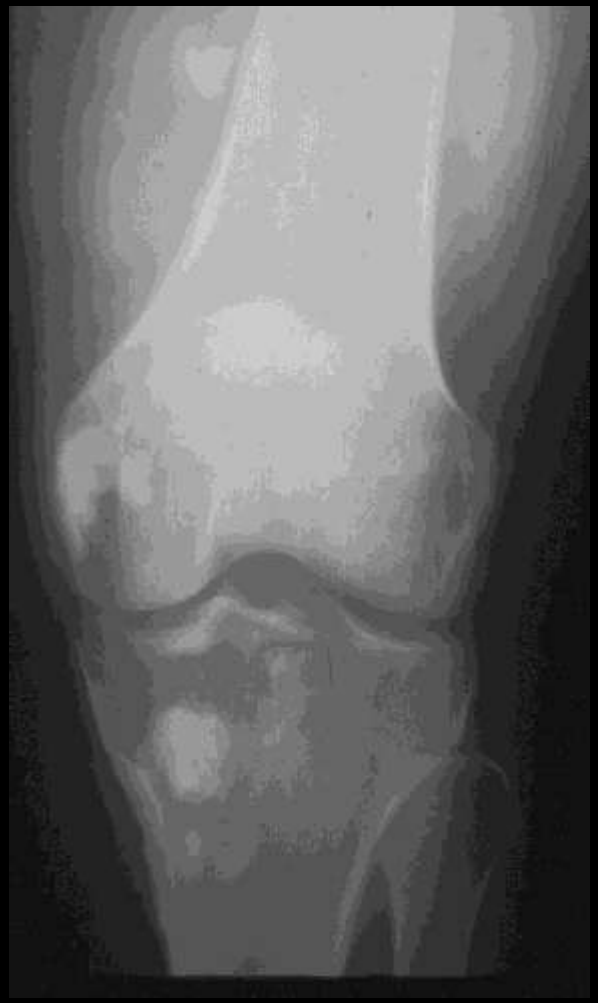
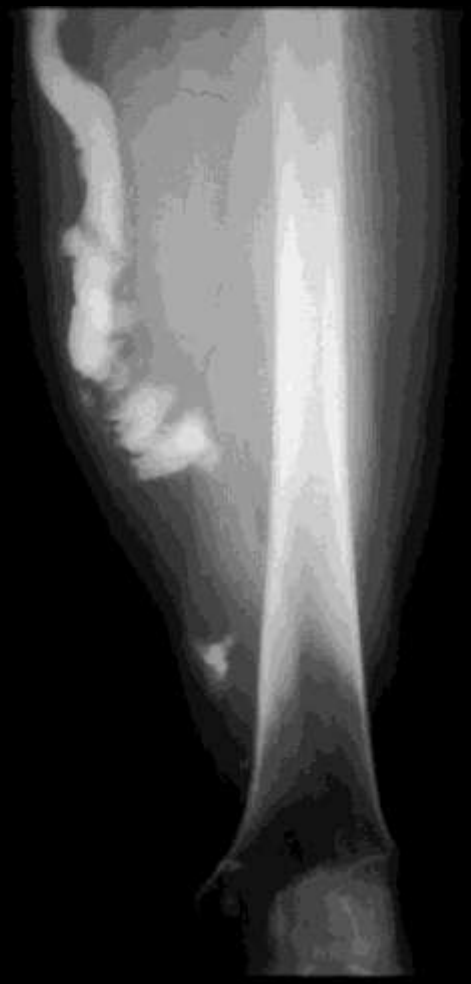
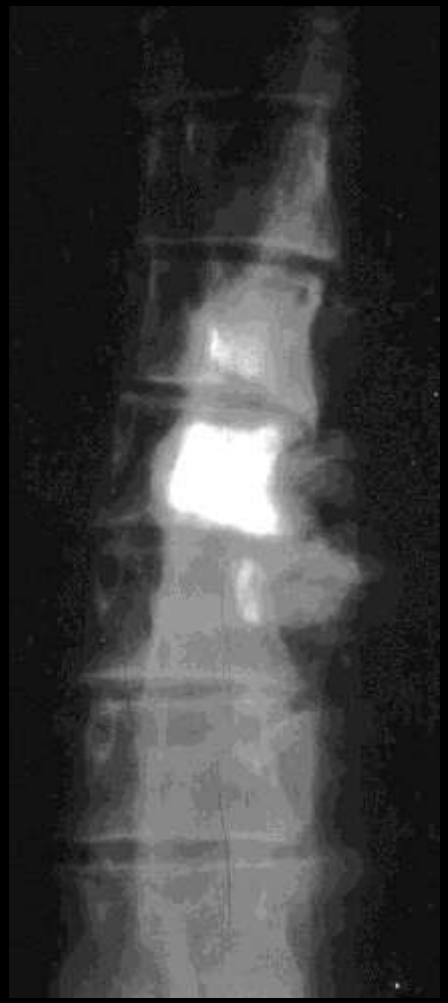
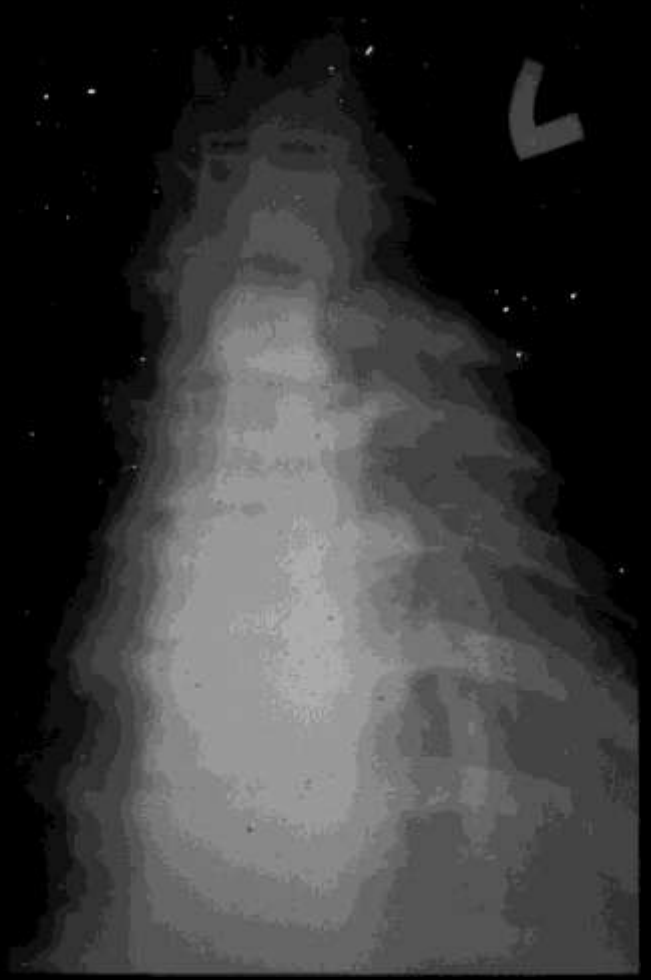




Melorrheostosis Meningocele Lipoma





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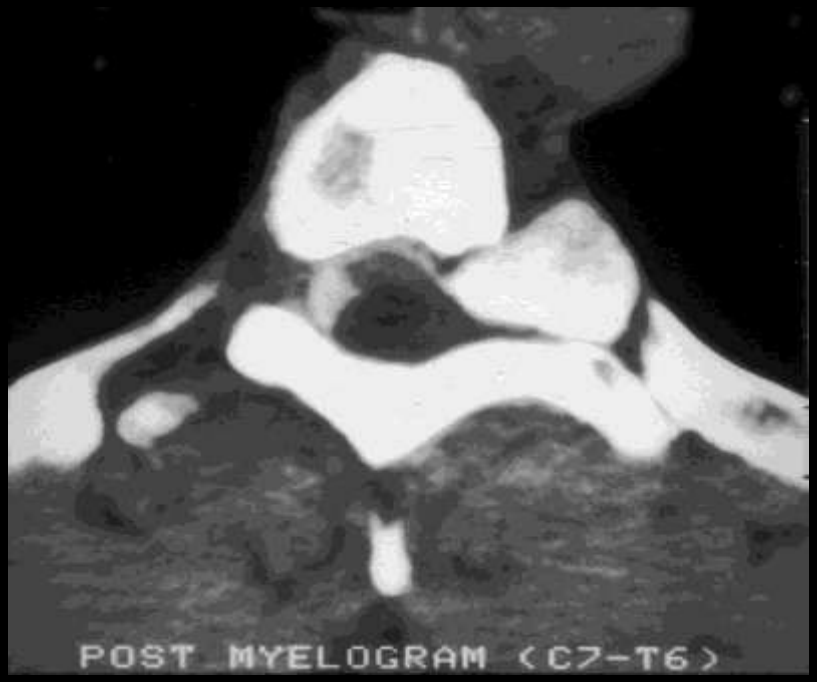
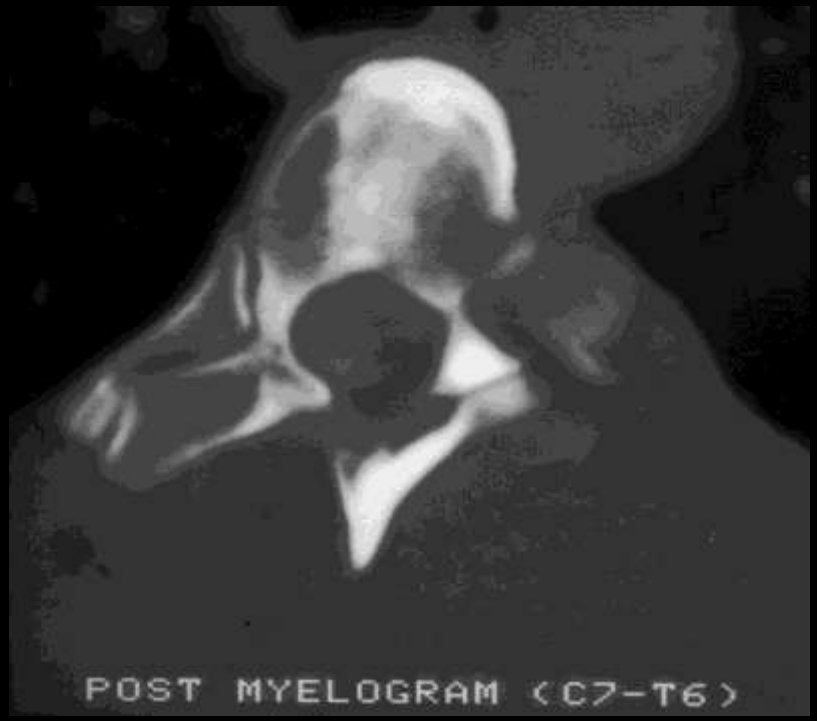
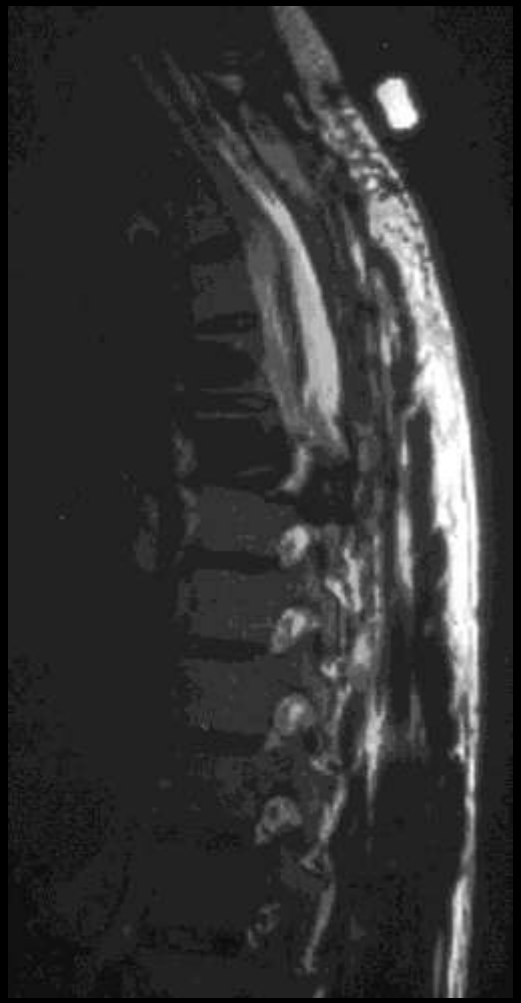




Melorrheostosis Meningocele Lipoma



Sag T2

22M

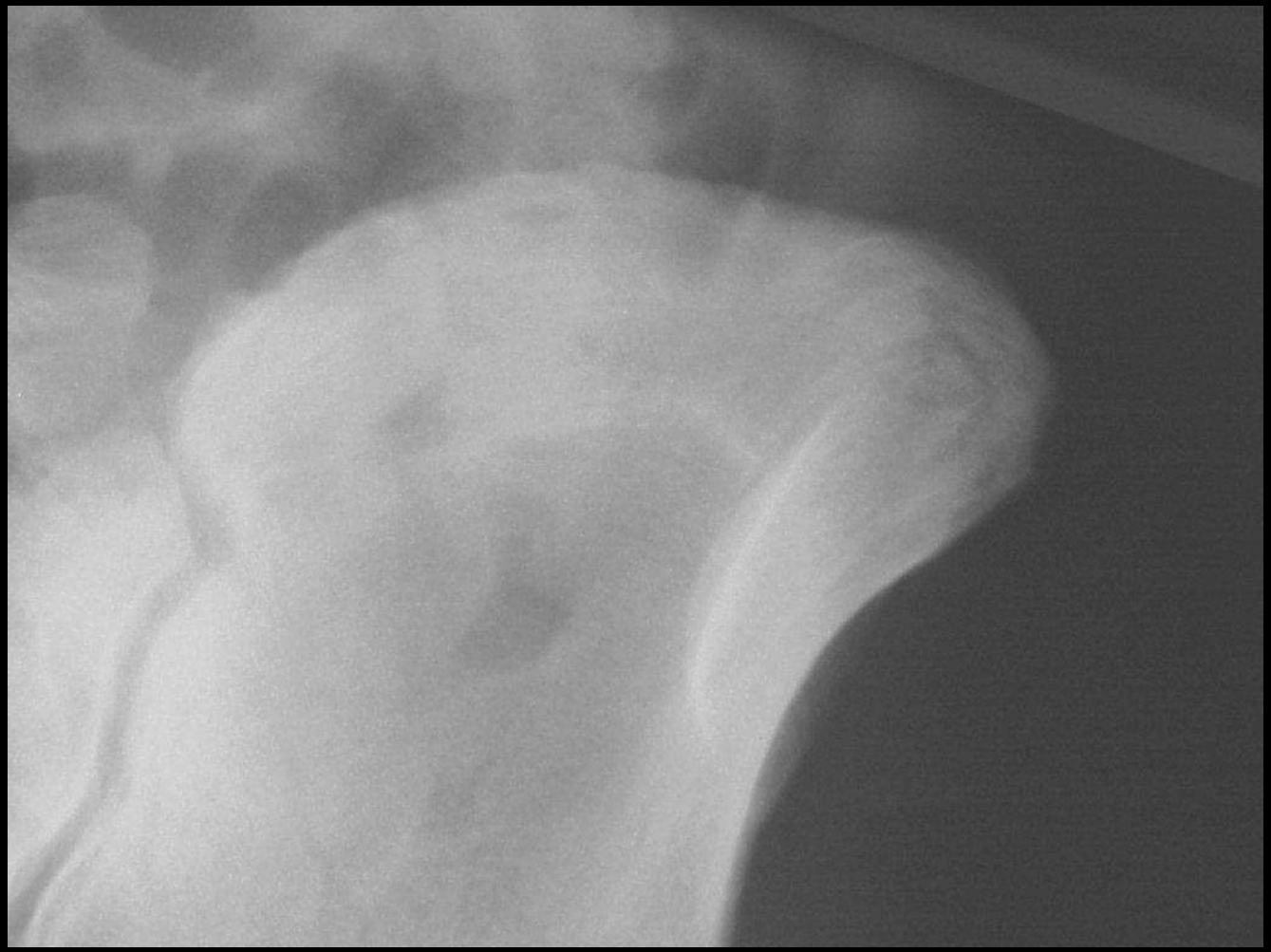
Sclerosis with Periosteal Reaction

- Traumatic
  - Healing fracture
- Neoplastic
  - Metastases
  - Lymphoma
  - Osteoid osteoma
  - Ewings
  - Chondrosarcoma
- Infective
  - Osteomyelitis
  - Syphilis
- Idiopathic
  - Infantile cortical hyperostosis
  - Melorrheostosis

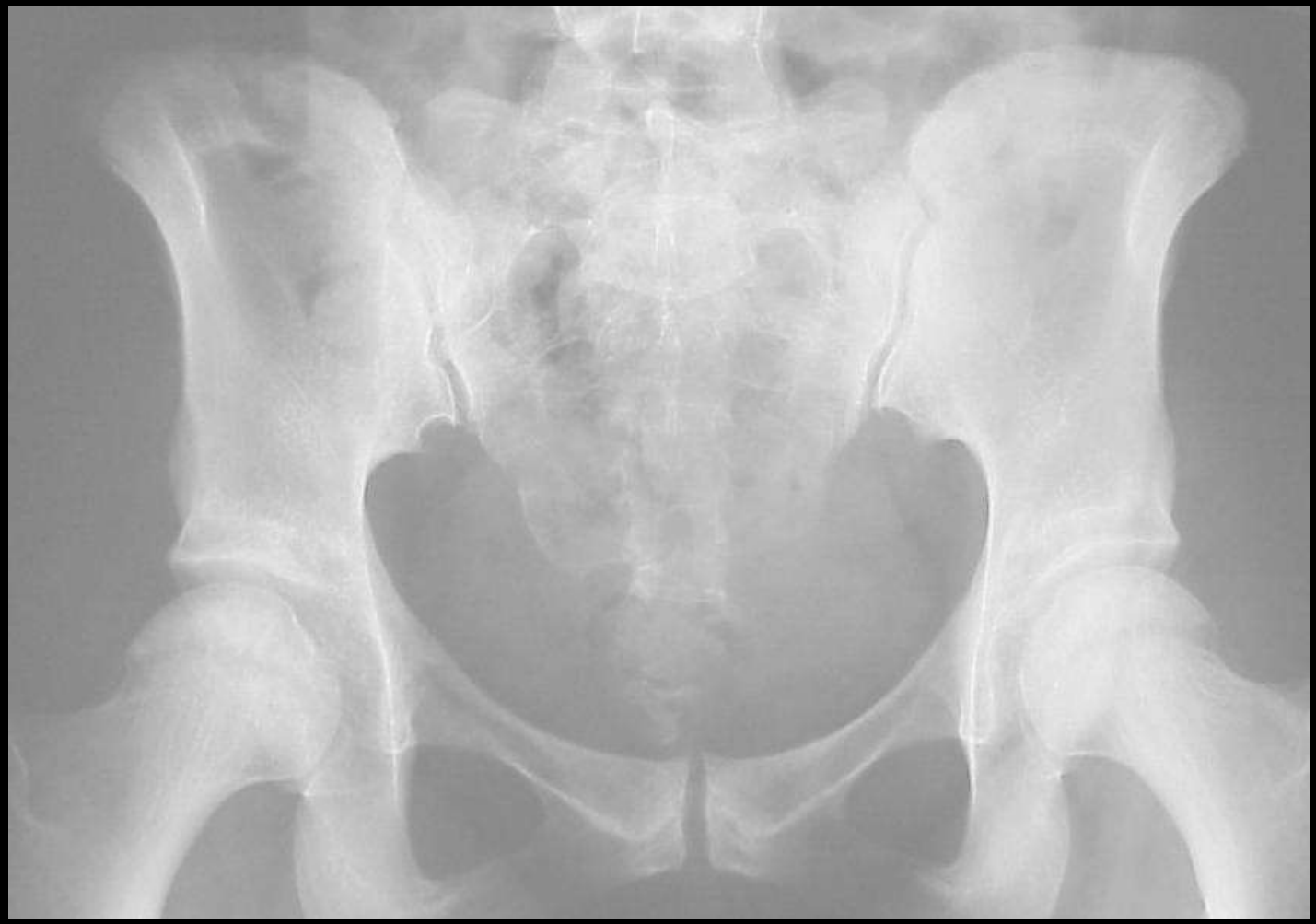
Osteopathia Striata (Voorhoeve's)

- Linear bands
- Parallel to long axis of bone
- Appendicular and Pelvis
- Skull and clavicles sparred

Osteopathia Striata (Voorhoeve's)



Osteopathia Striata (Voorhoeve's)



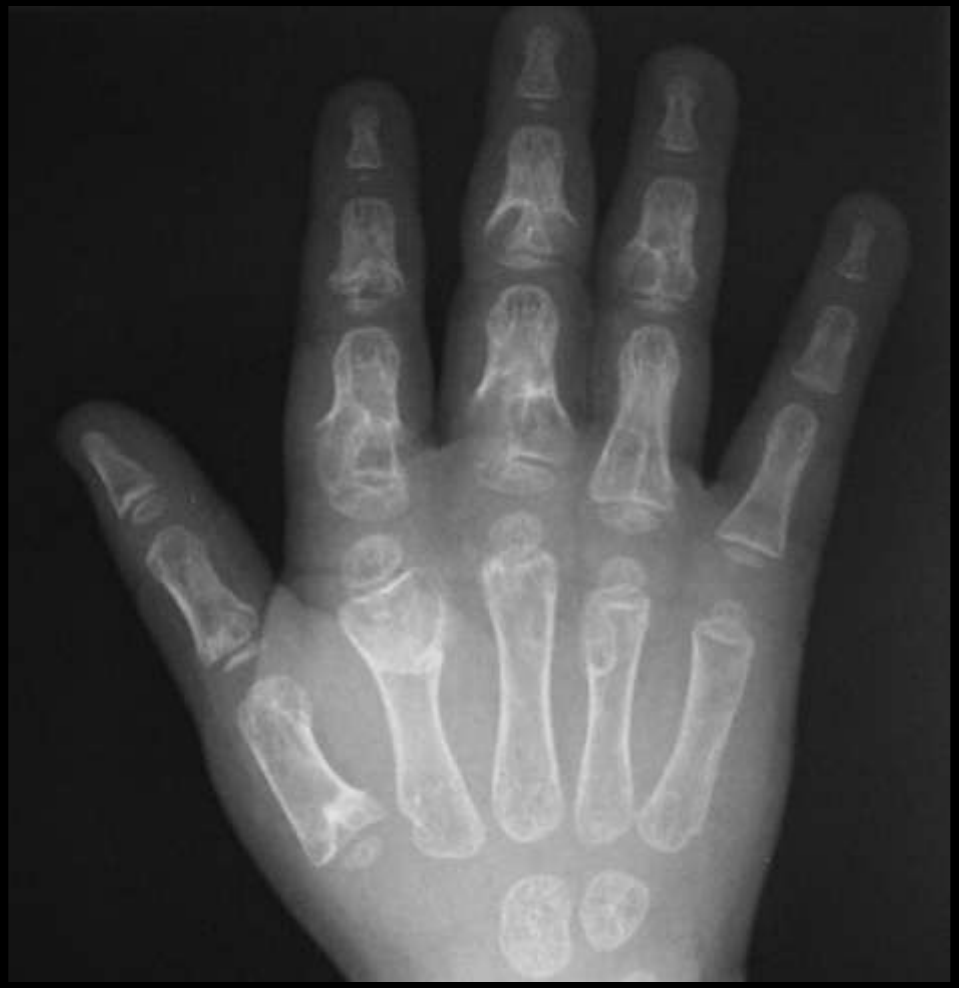
Dense Vertical Metaphyseal Lines

- Congenital Rubella
- Osteopathia Striata
- Hypophosphatasia
- Localized metaphyseal injury
- Enchondromatosis

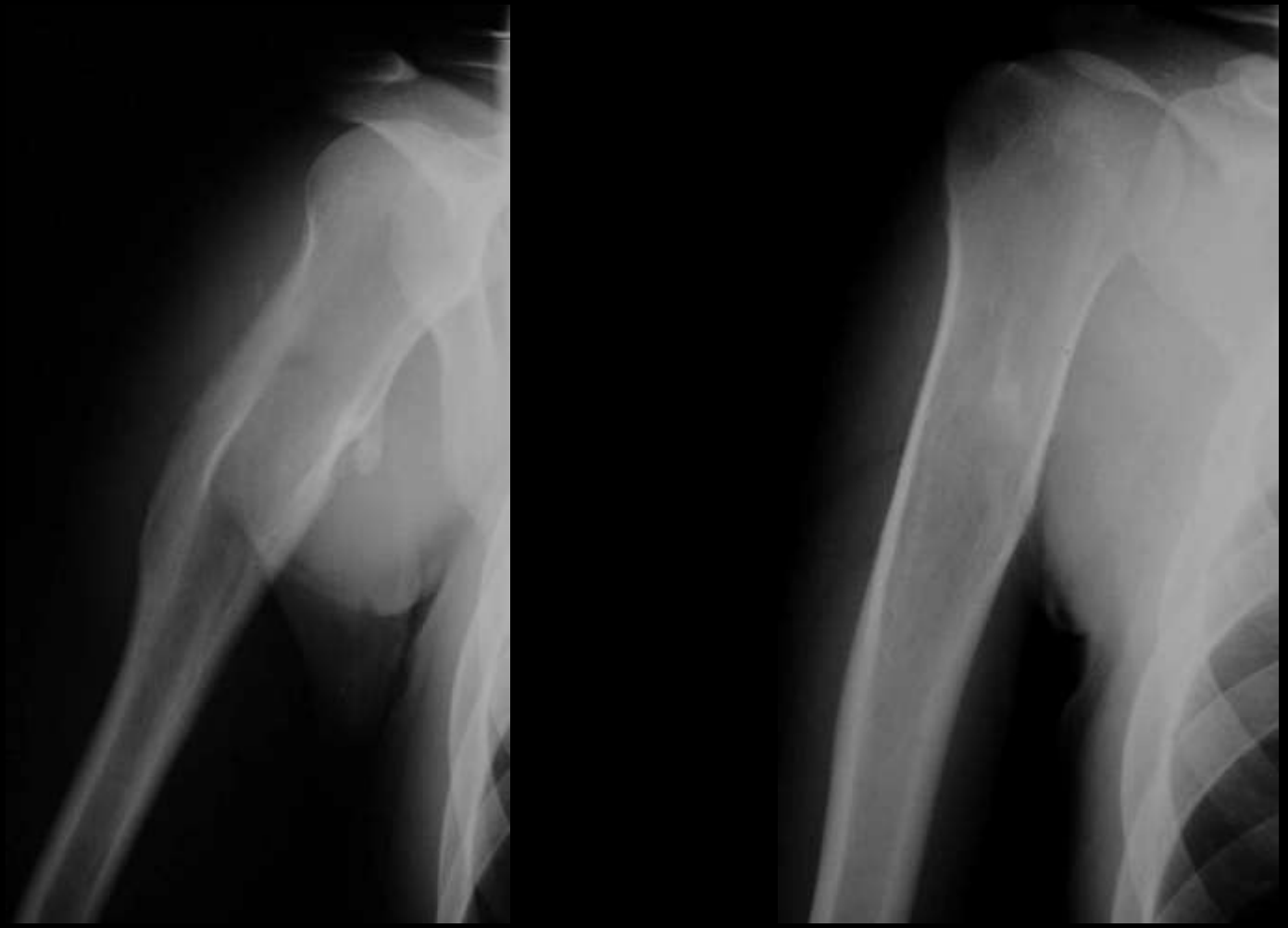
Ollier's Syndrome

- Multiple enchondromata
- Metaphyses and Diaphyses
- Multiple bones
- Not hereditary
- Appears in childhood
- Tends to be unilateral
- Vertical striated metaphyses
- Growth deformities
- Chondrosarcoma 25%

Ollier's Syndrome



Olliers Disease



24M

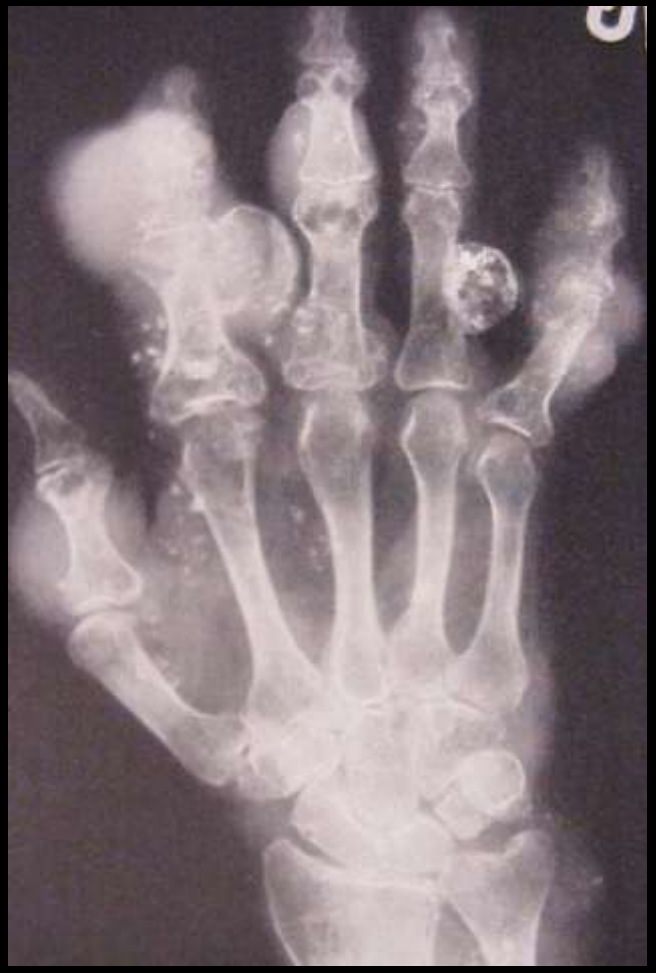
Medullary Lucency with Thin Sclerotic Border

- Geode
- Healing benign or malignant lesion
- Brodie's abscess
- Benign bone neoplasm
  - Simple bone cyst
  - Enchondroma
  - Chondroblastoma
- Fibrous dysplasia

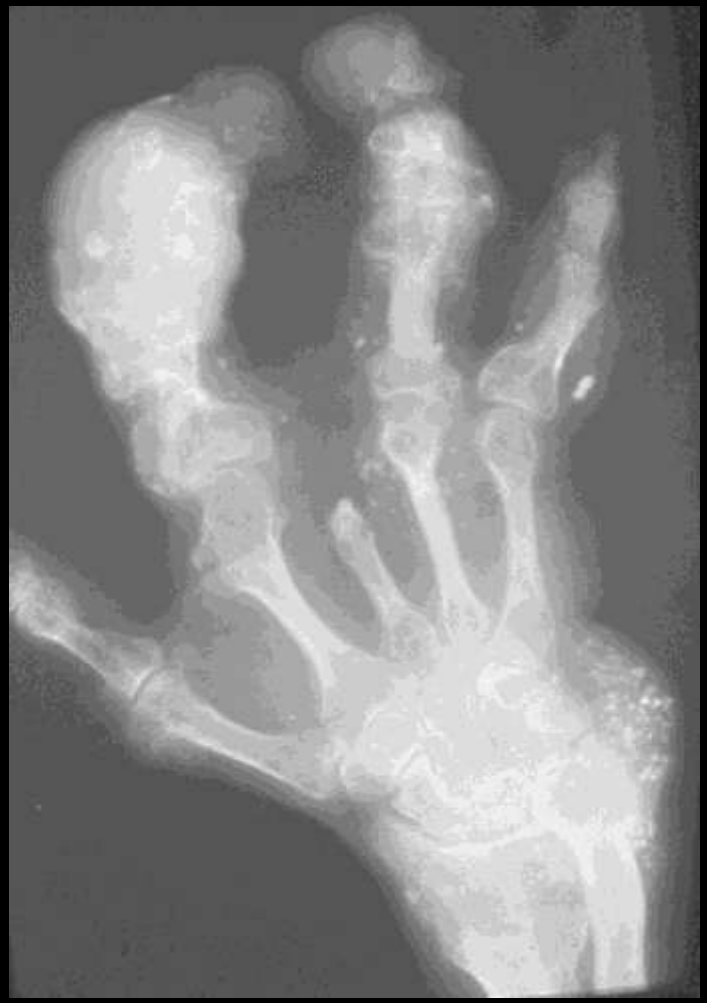
Maffucci's Syndrome

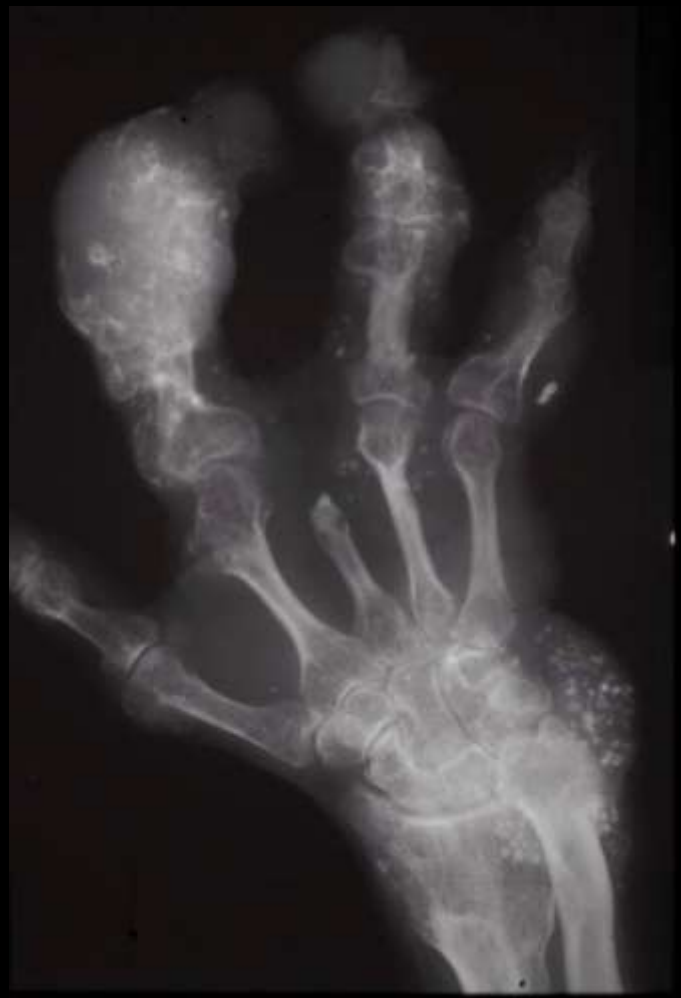
- Enchondromatosis
- With ST hemangiomas
- Phleboliths
- Malignant transformation 100%

Maffucci's Syndrome



Maffucci's Syndrome





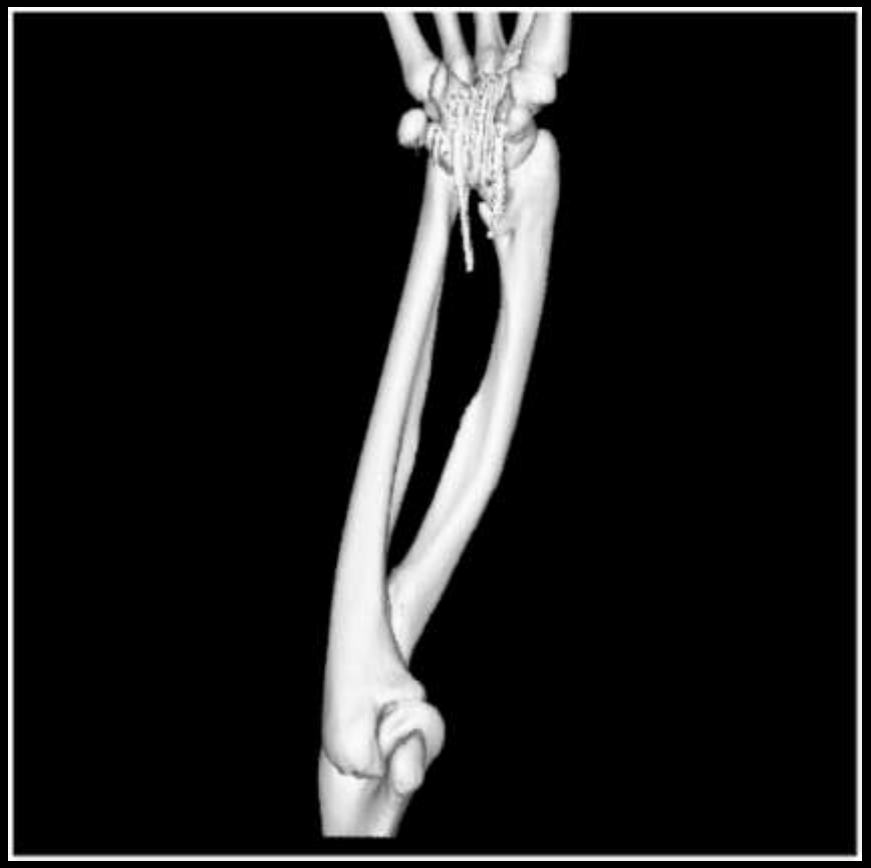
Expansile Lucent Lesion

- Primary Malignant Bone Neoplasms
  - Plasmacytoma
  - Chondrosarcoma
  - Telangiectatic osteosarcoma
- Secondary Malignant Bone Neoplasm
  - Mets
    - Thyroid, Renal, Breast, Lung, Melanoma, Phaeo
- Benign Bone Neoplasms
  - ABC
  - GCT
  - Enchondroma
- Non Neoplastic
  - Fibrous Dysplasia
  - Hemophilic pseudotumor
  - Brown tumor
  - Hydatid

Madelung Deformity

- Radial bowing of radius
- Dorsal bowing of radius
- Posterior dislocation of distal ulna
- Steep carpal angle
- Volar and ulna angled distal radius

Madelung Deformity



Madelung Deformity - Causes

- Isolated
  - Bilateral F>M
- Dyschondrosteosis (Leri-Weil)
  - Bilateral with mesomelic limb shortening
- Multiple hereditary exostosis
  - (Diaphyseal aclasis)
- Turner syndrome
- Post traumatic
- Post infective

Bone Dysplasia Terminology

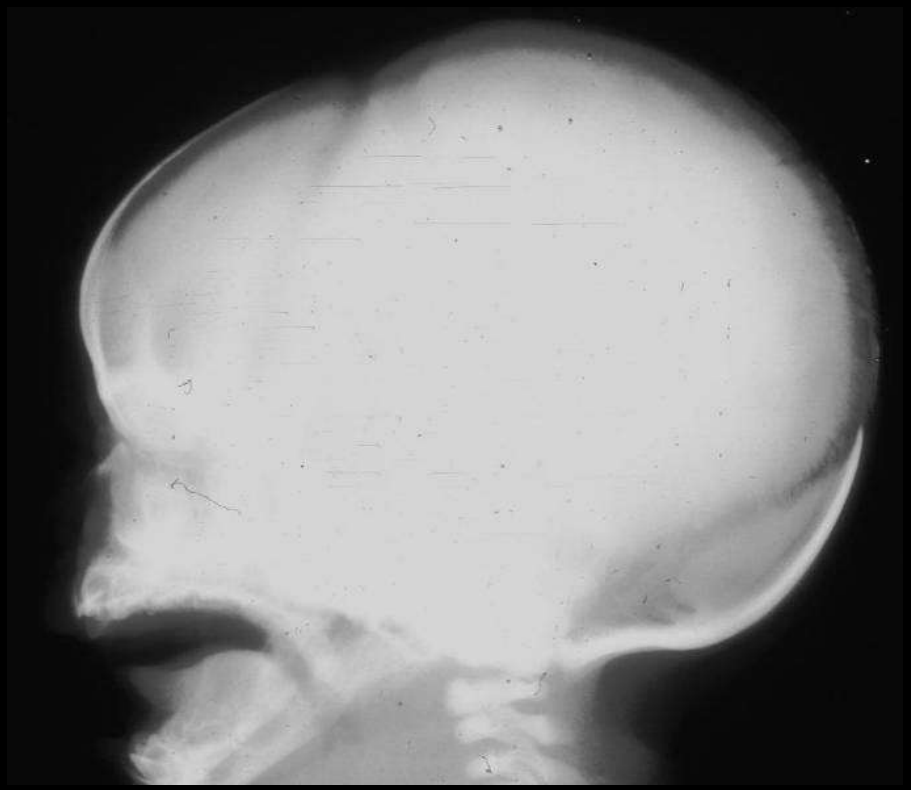
- Acro
  - Tip
- Meso
  - Middle – Dyschondrosteosis
- Acromesomelic
  - Middle and distal – Chondroectodermal dysplasia
- Rhizo
  - Root – Achondroplasia, Chondrodysplasia punctata
- Phocomelia
- Varus – Distal deviated towards midline
- Valgus – Distal deviated away from midline

Achondroplasia
General

- Defect of endochondral bone
- AD
- 80% spontaneous mutations

Achondroplasia
Radiology - Skull

- Large skull
- Small base of skull
- Small face
- Small sella
- Steep clivus
- Small foramen magnum
- Funnel foramen magnum
- Occipitalisation of C1



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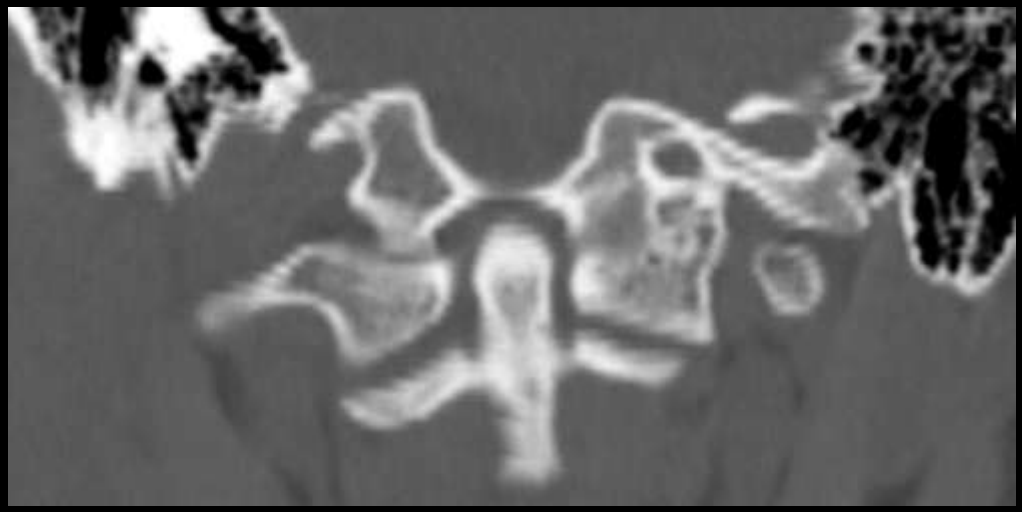
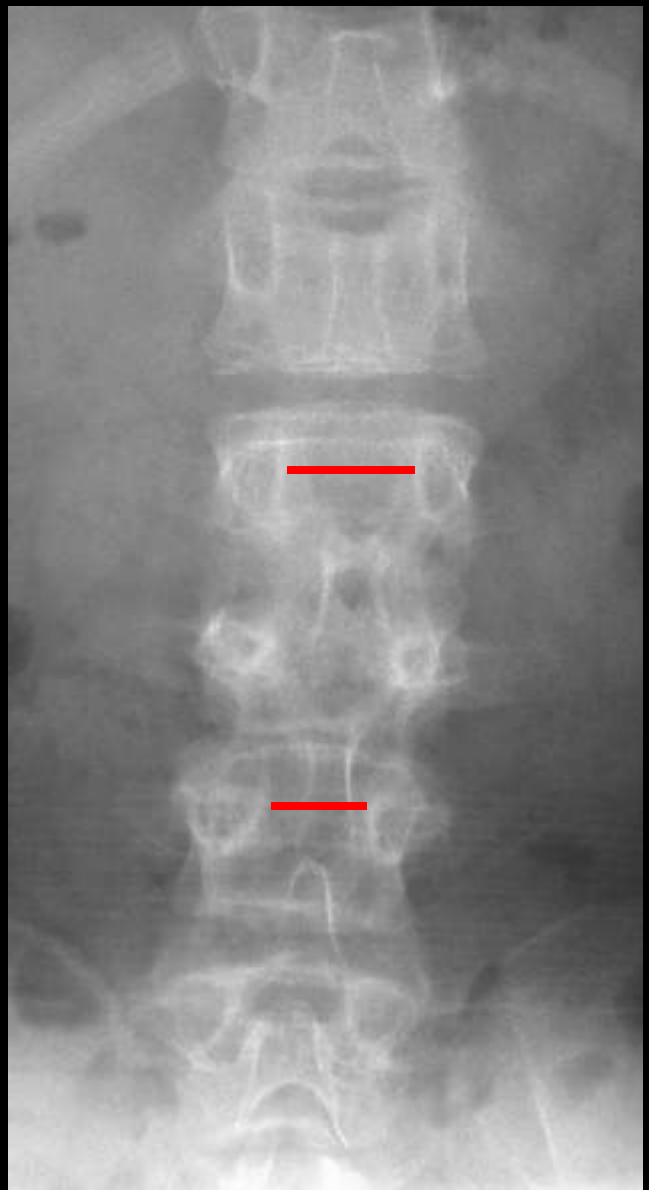


Achondroplasia
Radiology – Axial Skeleton

- Short ribs
- Concave ribs anteriorly
- Stubby sternum
- Decreasing interpediculate distance inferiorly
- Short pedicles
- Posterior vertebral scalloping
- Lumbar Kyphosis -> Lordosis
- Spinal stenosis
- Anterior vertebral body beak at T12/L1-2



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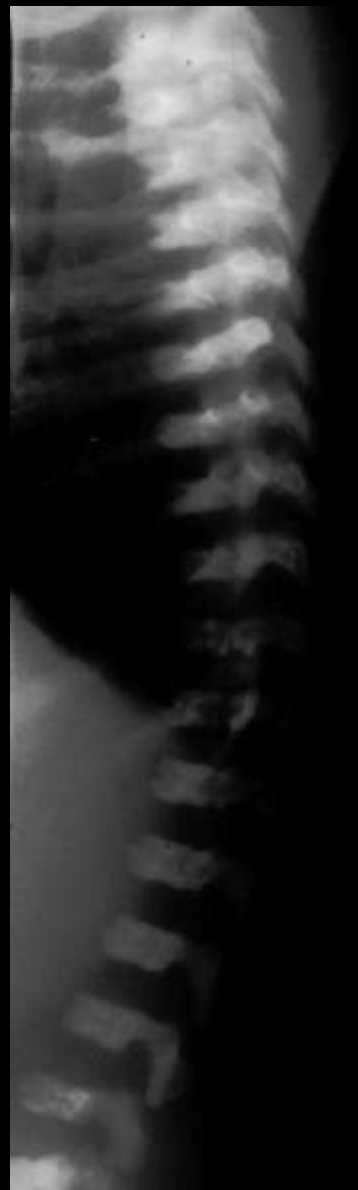

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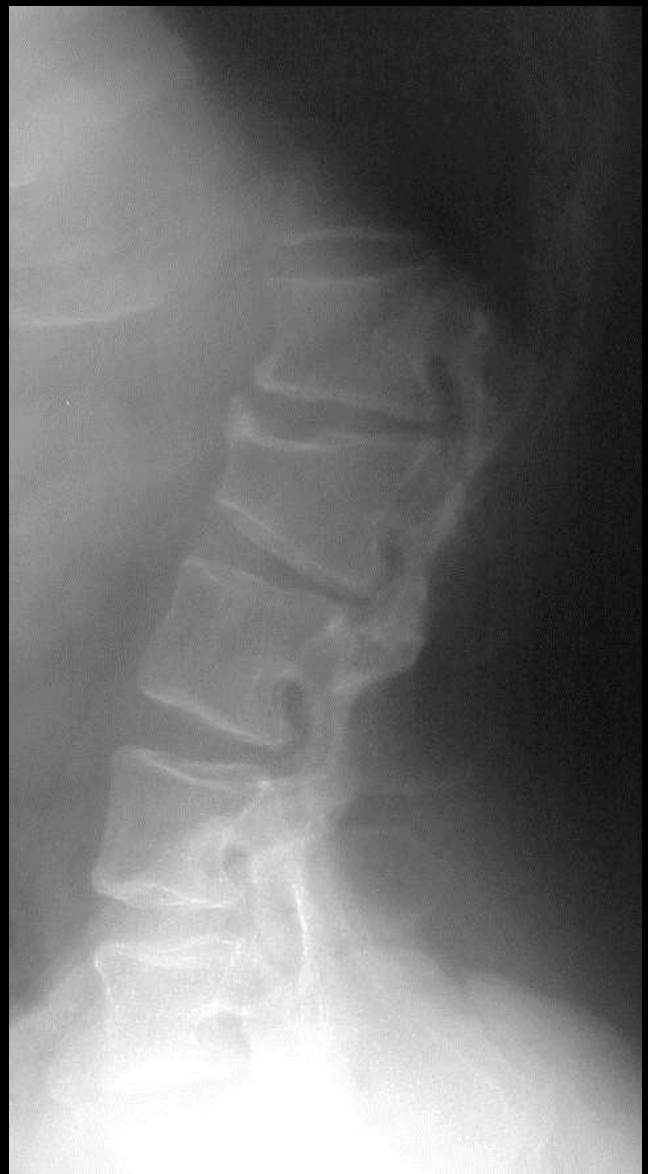
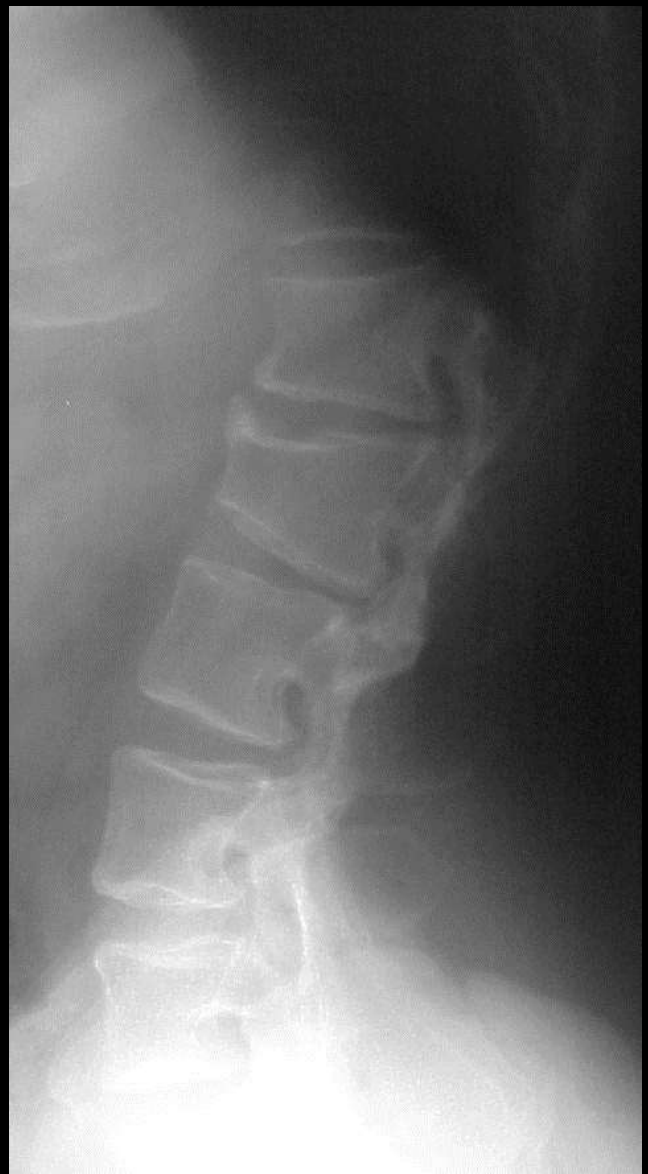


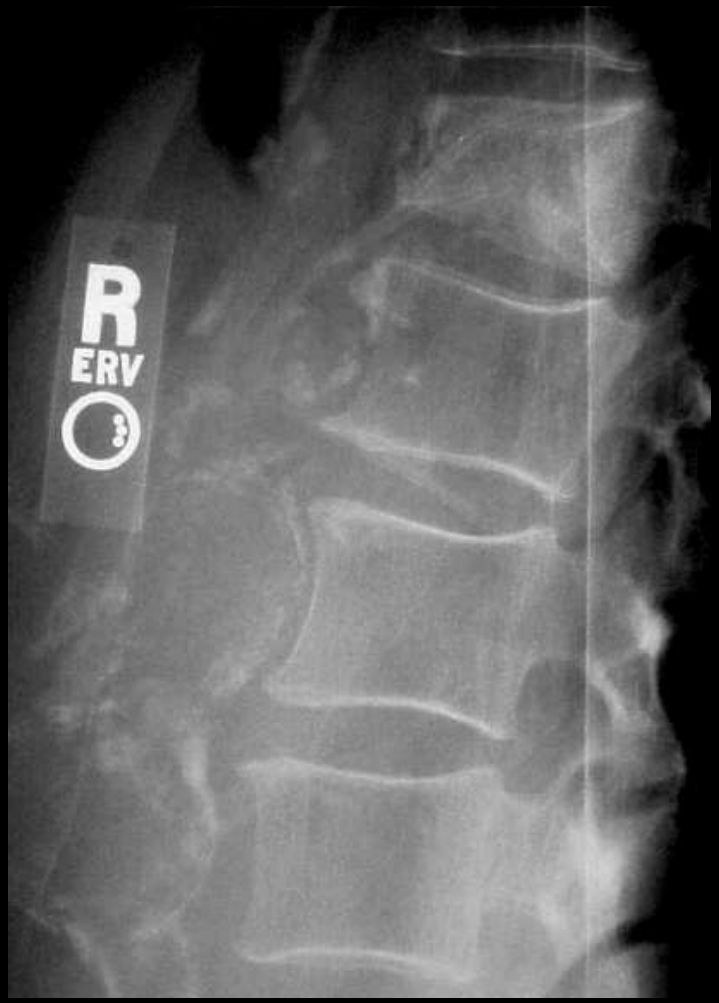
Posterior Vertebral Scalloping

- Tumors of spinal canal
- Neurofibromatosis
- Dural ectasia
- Acromegaly
- Achondroplasia
- Communicating hydrocephalus
- Syringomyelia
- Congenital syndromes
  - Ehlers Danlos
  - Marfan’s
  - Hurler’s
  - Morquio’s
  - Osteogenesis imperfecta



Anterior Vertebral Scalloping

- AAA
- Lymphadenopathy



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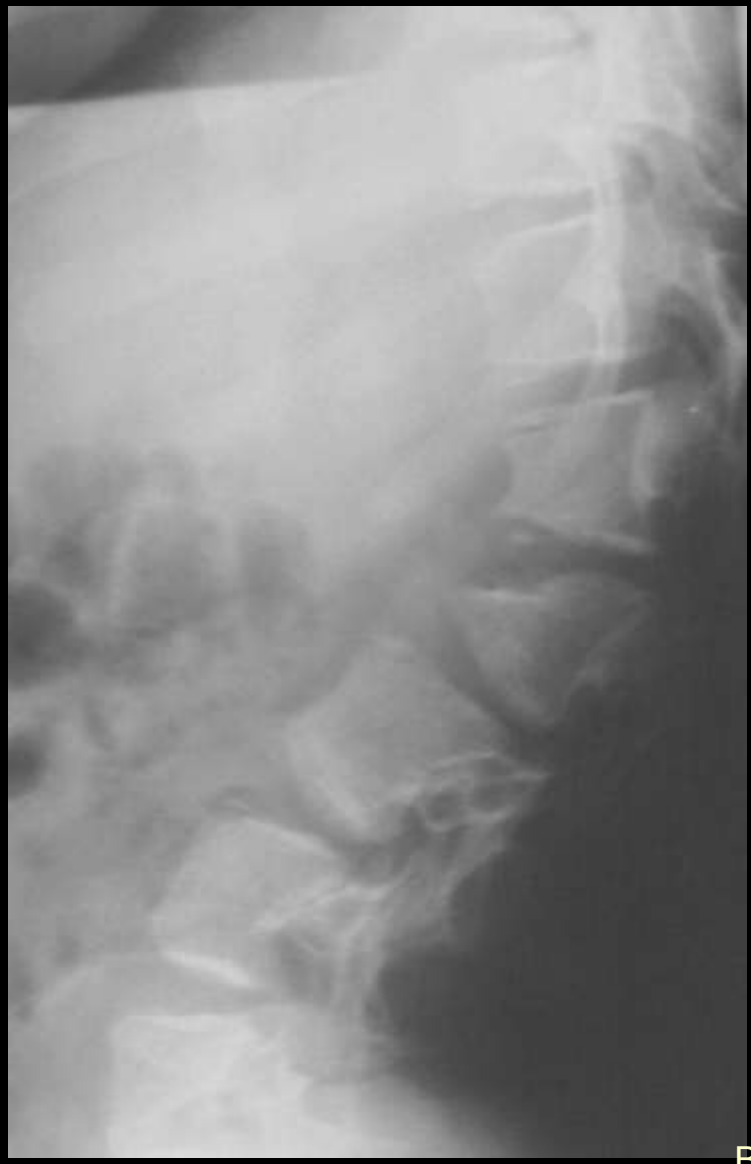
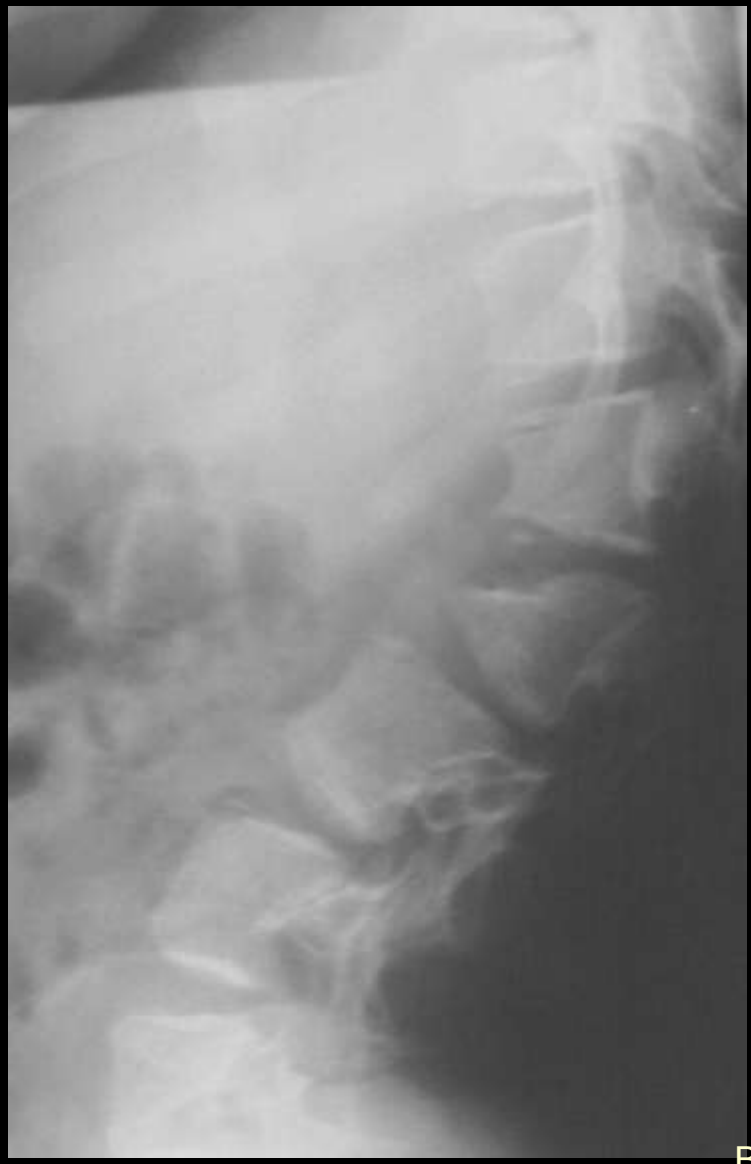


Bullet

Anterior vertebral body beak at T12/L1-2

- Central
  - Morquio,s
- Inferior
  - Hurler’s
  - Achondroplasia
  - Pseudoachondroplasia
  - Cretinism
  - Down’s syndrome
  - Neuromuscular disorders



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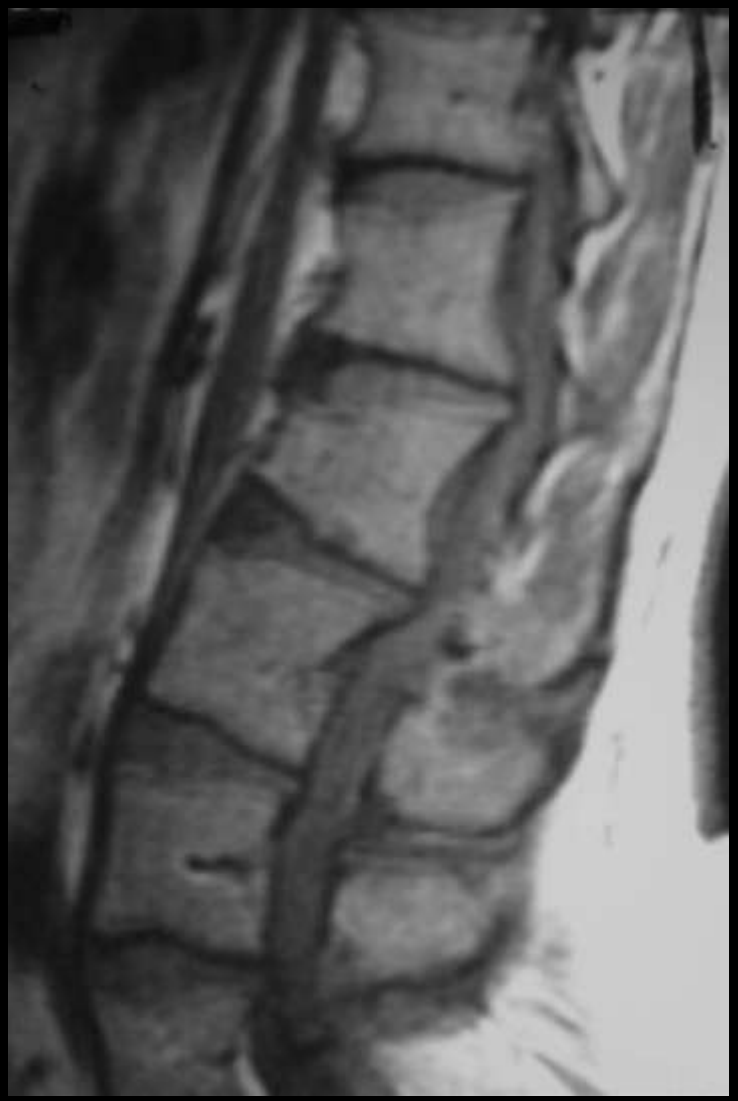
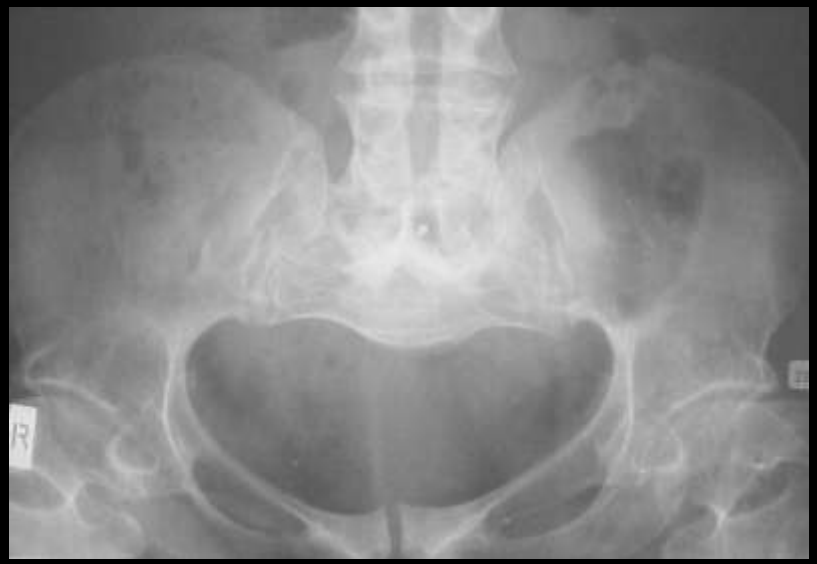


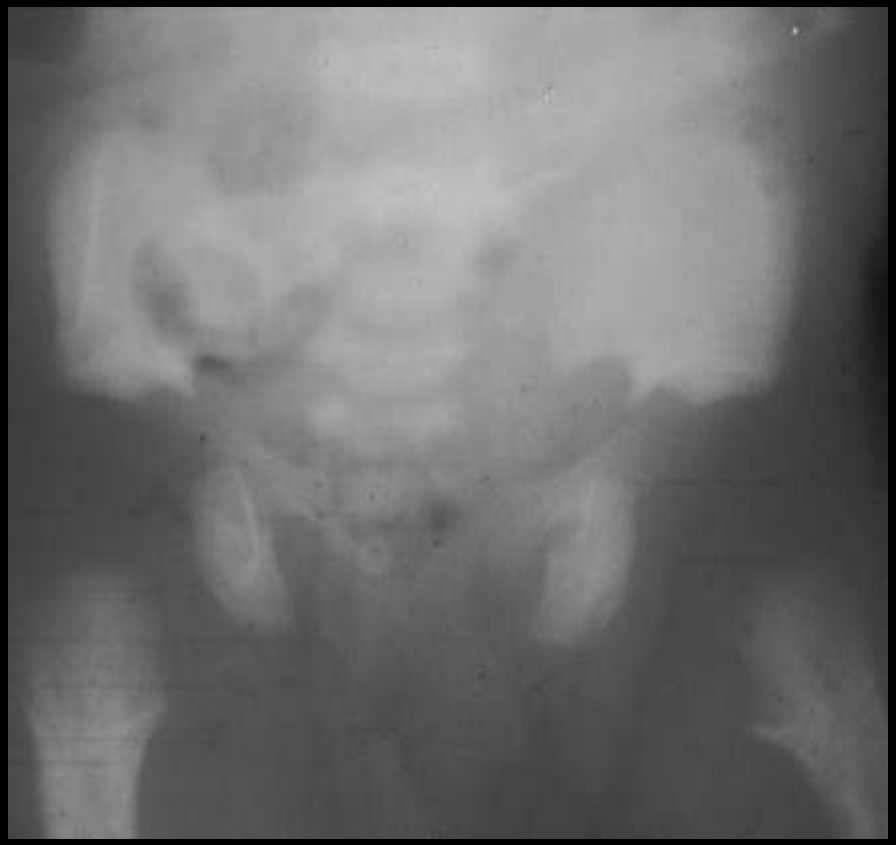
Achondroplasia
Radiology – Axial Skeleton

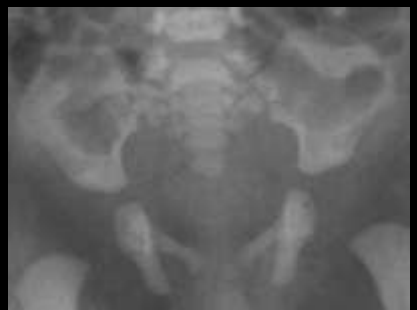
- Short ribs
- Concave ribs anteriorly
- Stubby sternum
- Decreasing interpediculate distance inferiorly
- Short pedicles
- Posterior vertebral scalloping
- Lumbar Kyphosis -> Lordosis
- Spinal stenosis
- Anterior vertebral body beak at T12/L1-2



Achondroplasia
Radiology – Pelvis

- Champagne glass pelvis
- Square iliac bones
- Horizontal acetabular roof
- Narrow greater sciatic notch
- Narrow pelvic inlet
- Low sacral articulation on ilia

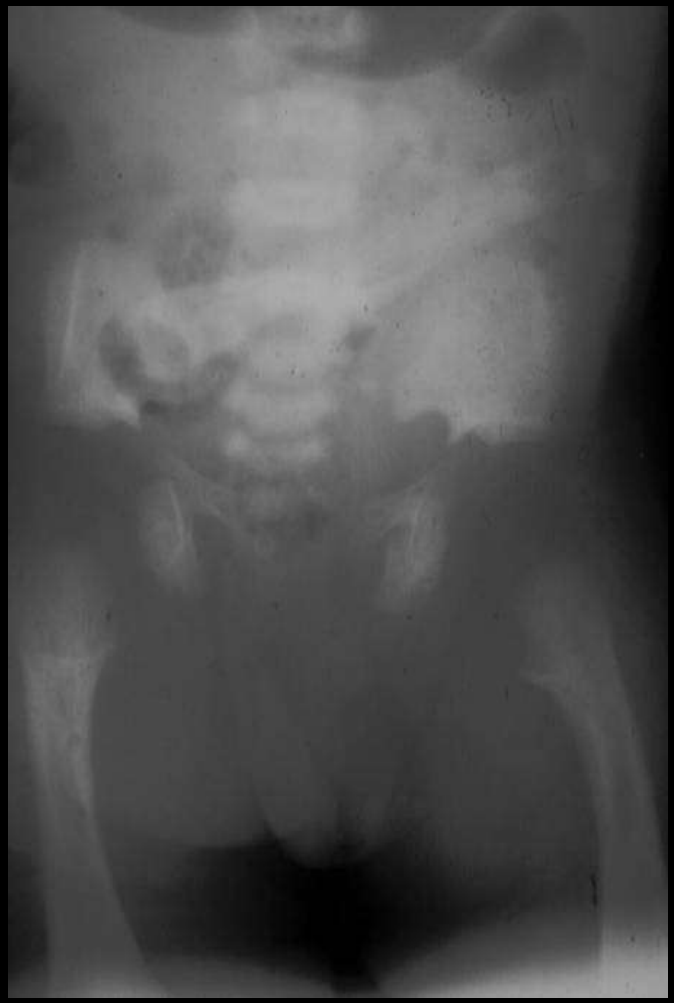




Newborn

Achondroplasia
Radiology – Appendicular Skeleton

- Rhizomelic micromelic with bowing
- Wide chevron metaphyses
- Ball and socket epimetaphyseal junction
- Flared metaphyses



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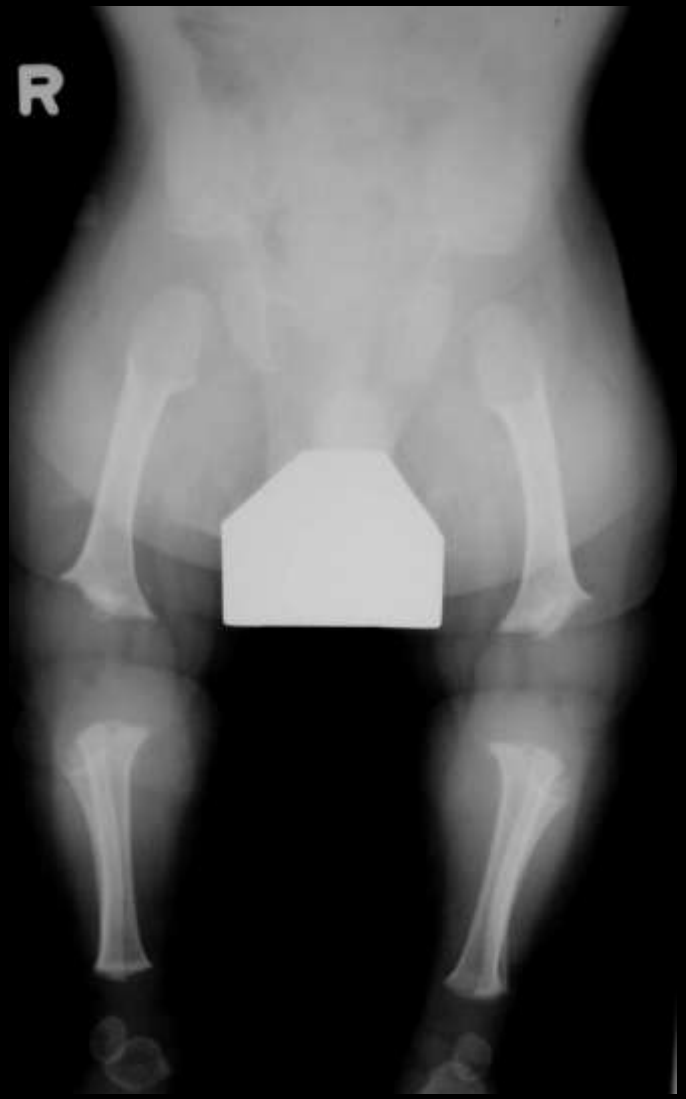


Metaphyseal Cupping

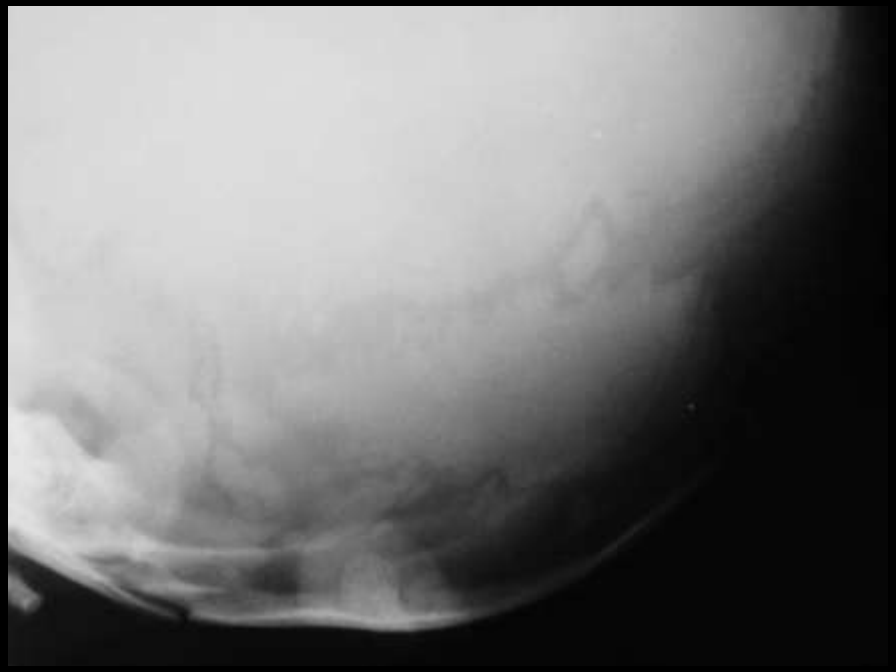
- Normal
  - Distal ulna, proximal fibula
- Rickets
  - With widening and fraying
- Trauma
- Bone Dysplasia
  - Achondroplasia
  - Pseudoachondroplasia
  - Metatropic dwarfism
  - Diastrophic dwarfism
  - Metaphyseal chondrodysplasia
  - Hypophosphatasia
- Scurvy

Cleidocranial Dysostosis

- Autosomal Dominant
- 1/3 new mutations
- Retarded development of membranous bones

Cleidocranial Dysostosis

- Brachycephaly
- Wormian bones
- Frontal and Parietal bossing
- Wide sutures and fontanelles with delayed closure
- Broad mandible, small facial bones
- Delayed eruption and supernumary teeth
- Basilar invagination
  - Bulging of Cx spine and foramen magnum into skull
- Platybasia
  - angle between sphenoid roof and clivus >150^0



Wormian Bones

- CCD
- PKD
- Hypophosphatasia
- Osteogenesis Imperfecta
- Downs
- Cretinism
- Acro-osteolysis of Hajdu and Cheney
- Pachydermoperiostosis
- Menkes kinky hair syndrome

Cleidocranial Dysostosis

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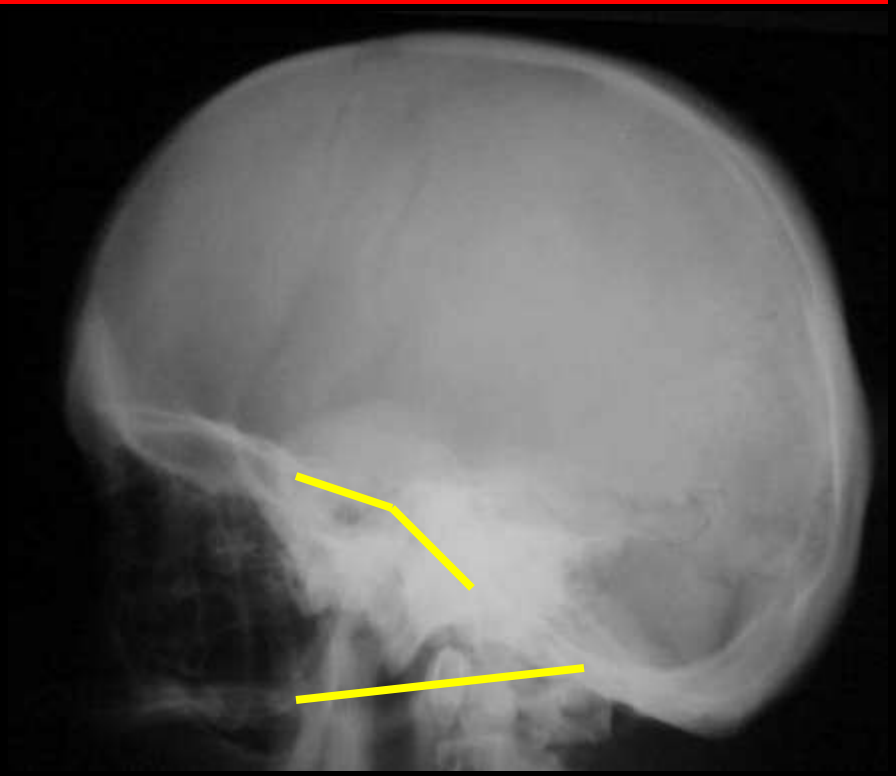

Chamberlains line

Sphenoid angle

Cleidocranial Dysostosis

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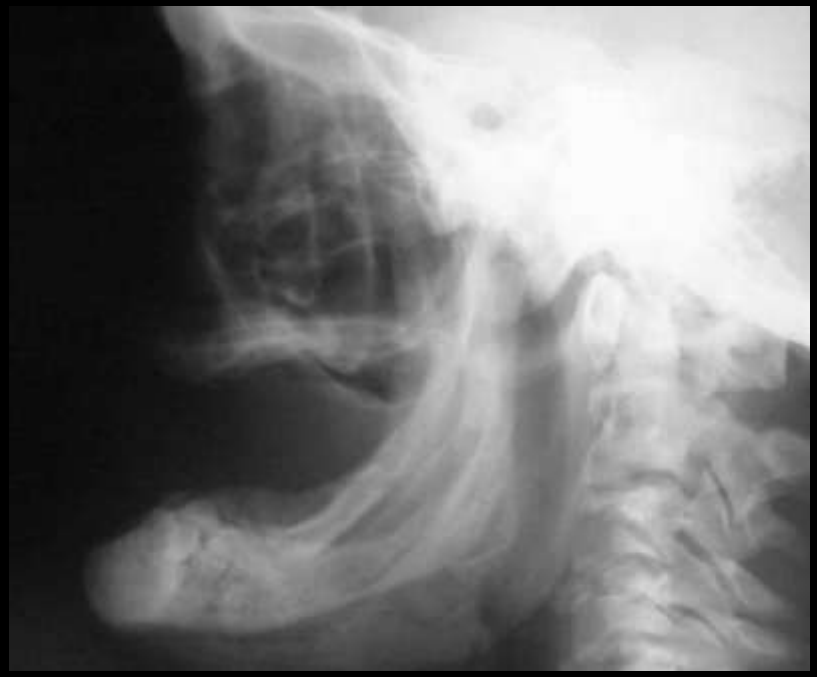
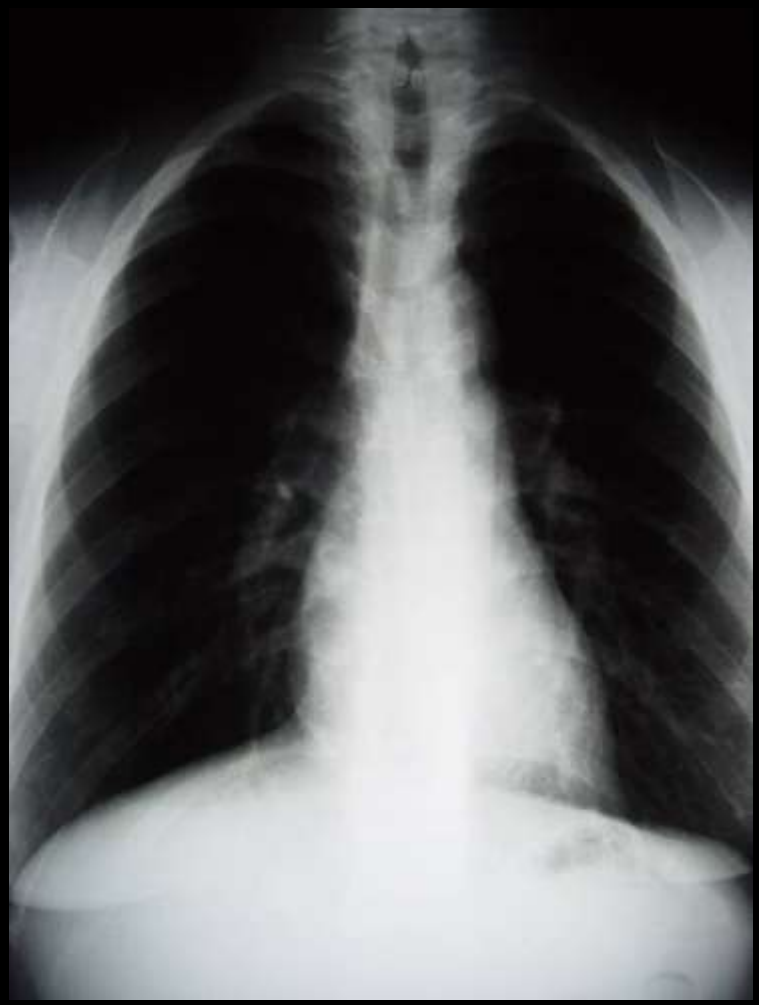


Bone Dysplasia Terminology

- Brachycephaly
  - Short wide skull
  - Premature closure of coronal suture
- Turricephaly / Oxycephaly / Acrocephaly
  - Turret shaped head (cone head)
  - Premature closure of coronal and sphenofrontal sutures
  - Seen in the Acrocephalosyndactylys (Aperts, Crouzon, Pfeiffer)
- Dolicocephaly / Scaphocephaly
  - Long narrow skull
  - Premature closure of sagittal suture
- Trigonocephaly
  - Wedged narrow forehead, hypotelorism
  - Premature closure of metopic suture
- Plagiocephaly
  - Asymmetry of skull
- Triphyllocephaly
  - Cloverleaf skull (Kleeblattschadel)
  - Thanatophoric dwarfism

Cleidocranial Dysostosis – Axial skeleton

- Partial or total absence of the clavicle – bilateral
  - Lateral > Medial
- Clavicle pseudoarthrosis
- Delayed pubic ossification
- Varus or valgus femoral neck
- Small high scapulae
- Neonatal respiratory distress from thoracic deformity



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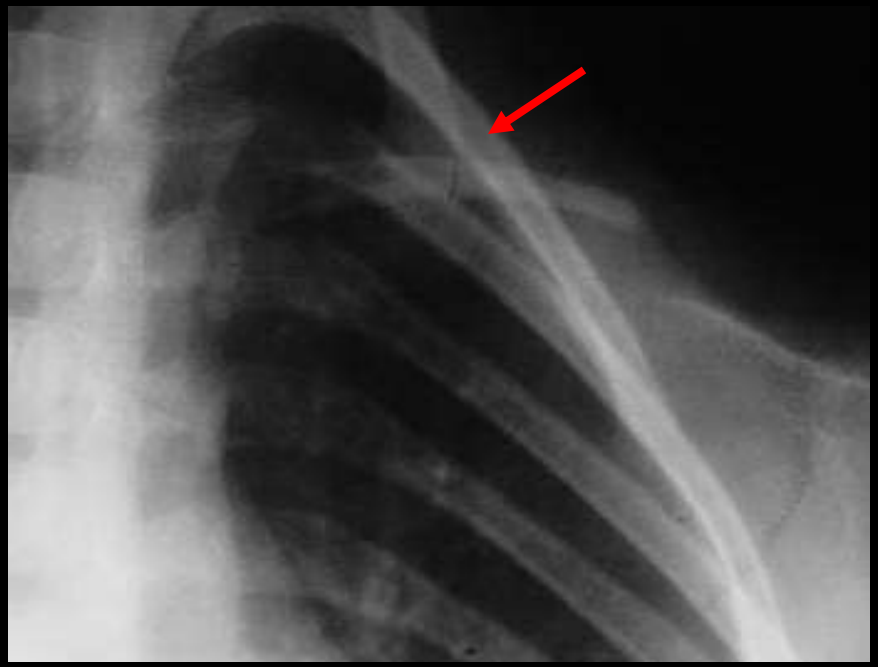

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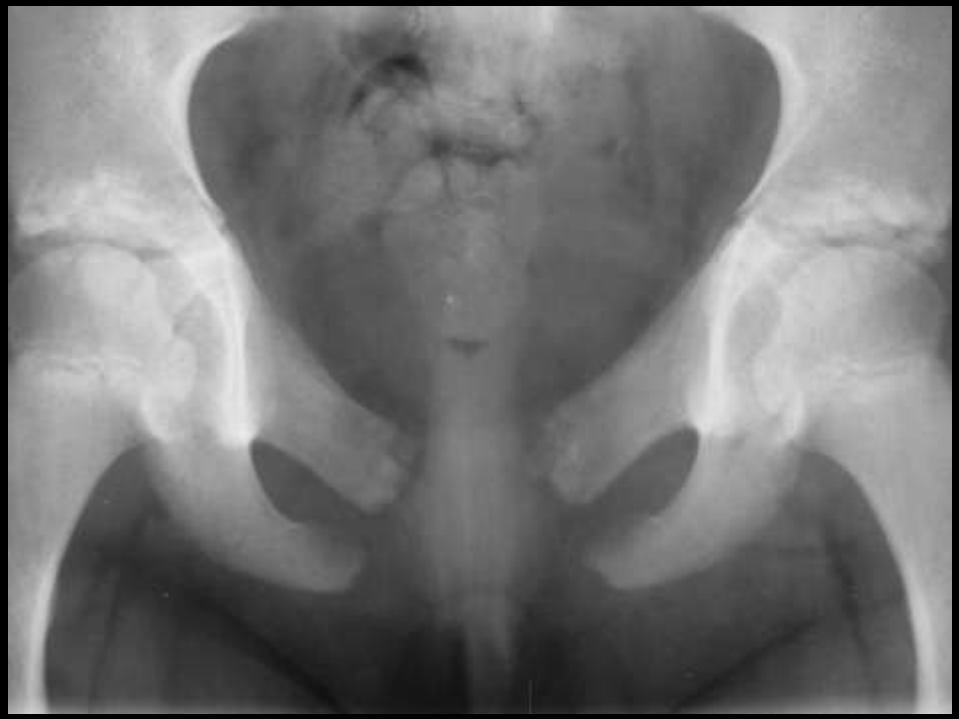
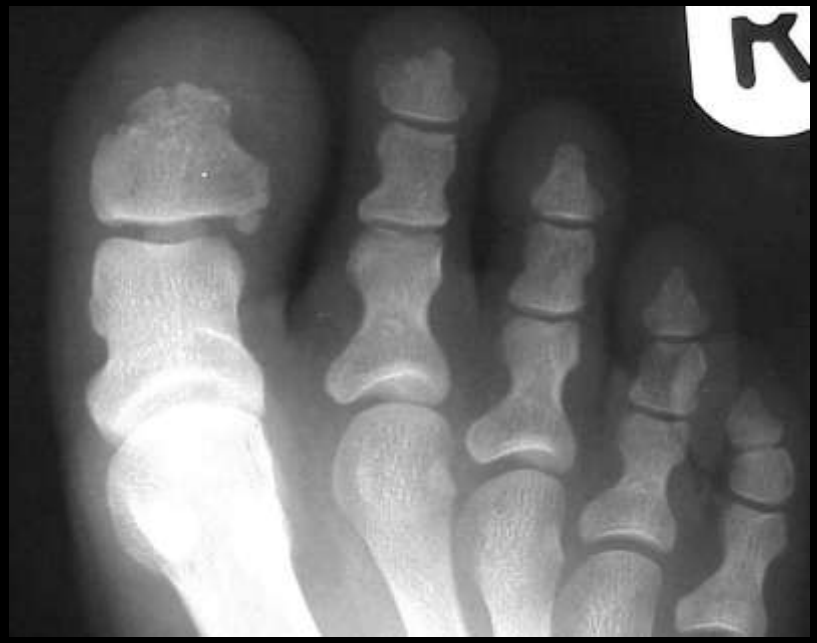


Wide pubic symphysis

- Trauma
- Cleidocranial dysostosis
- Bladder exstrophy
- Renal osteodystrophy
  - Amyloid
  - Hyperparathyroidism

Cleidocranial Dysostosis - Appendicular

- Long 2 +5 Metacarpals
- Short 2 + 5 Middle phalanges
- Cone shaped epiphyses
- Acroosteolysis
- Supernumerary ossification centers



Acroosteolysis

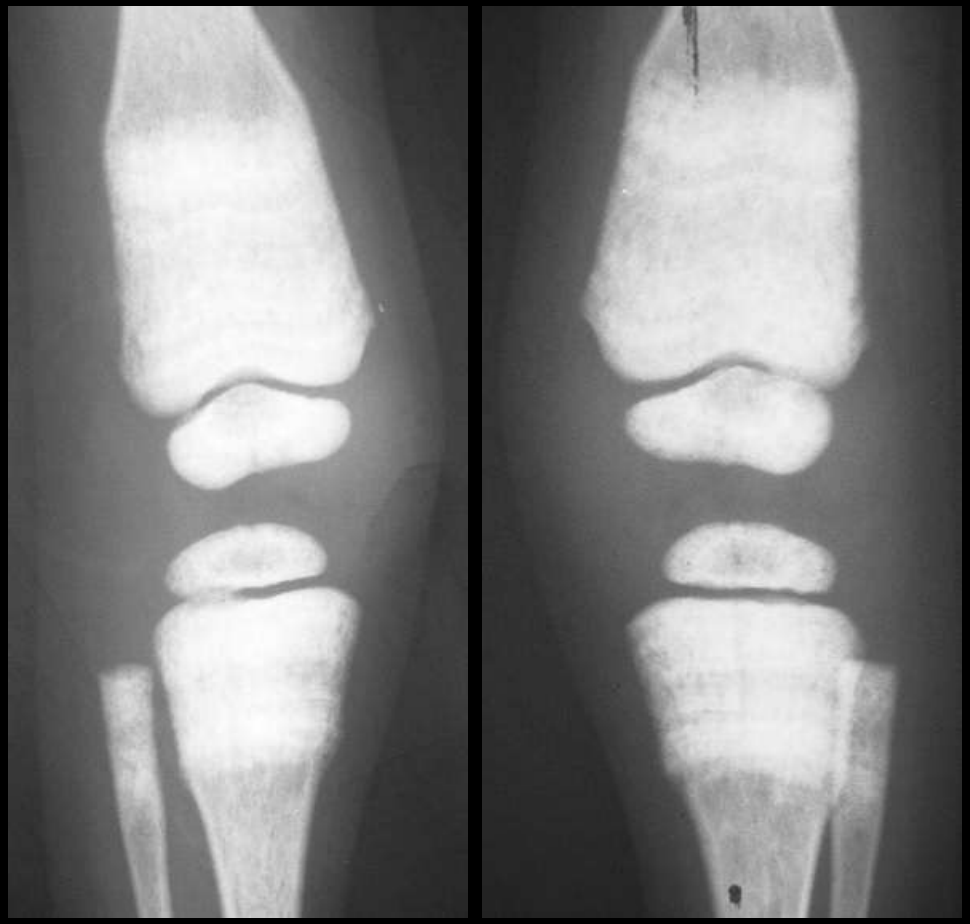
- Neuropathic
  - Syrinx, Myelomenigocele, Congenital insensitivity to pain, DM, Leprosy, Lesch Nyan syndrome
- Connective Tissue diseases
  - Scleroderma, Raynauds
- Trauma
  - Cold, Hot, Mechanical
- Hyperparathyroidism
- Psoriasis, Erosive OA, Multicentric reticulohistiocytosis
- Snake or scorpion venom
- Porphyria
- Idiopathic – Hajdu Cheney
- PVC

Osteopetrosis

- Marble bone disease
- Albers-Schonberg
- Defective osteoclasts
- Tarda
  - AD, Adult, Fxs, anemia, CN palsy
- Congenita
  - AR, Infantile, Systemic, Hepatosplenomegaly, Leukemia

Osteopetrosis

- Sclerotic, peri and endosteal
- Erlenmeyer flask.
- Bone in bone, sandwich vertebrae
- Rugger jersey spine
- Calvaria and mandible spared
- Transverse metaphyseal bands
- Fractures



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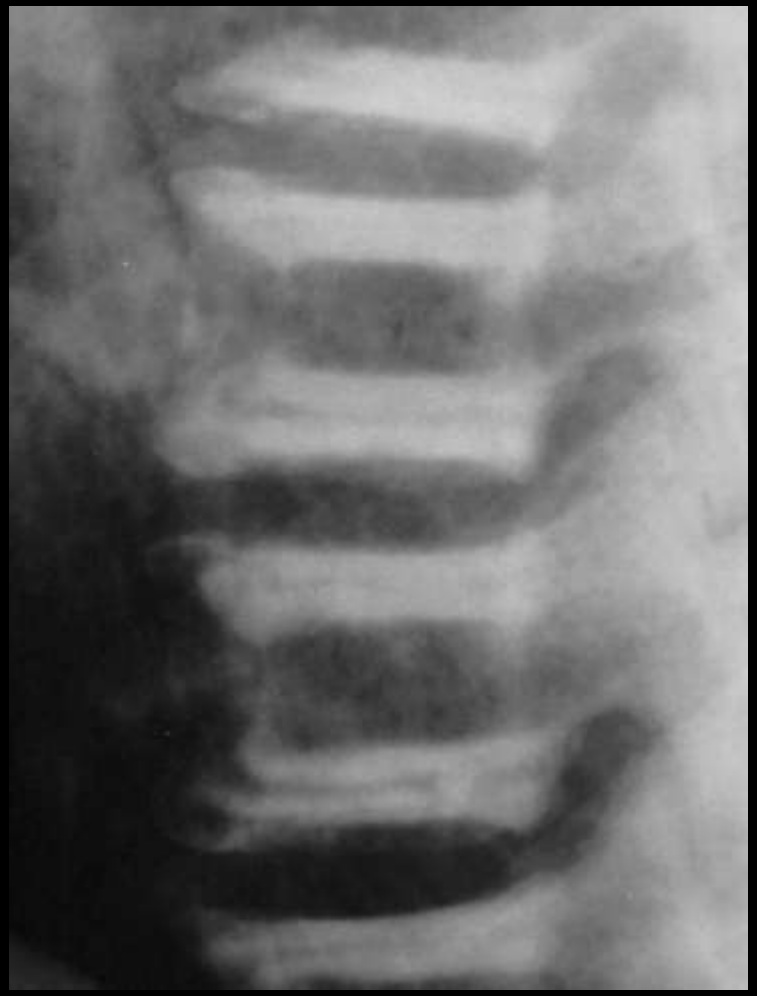
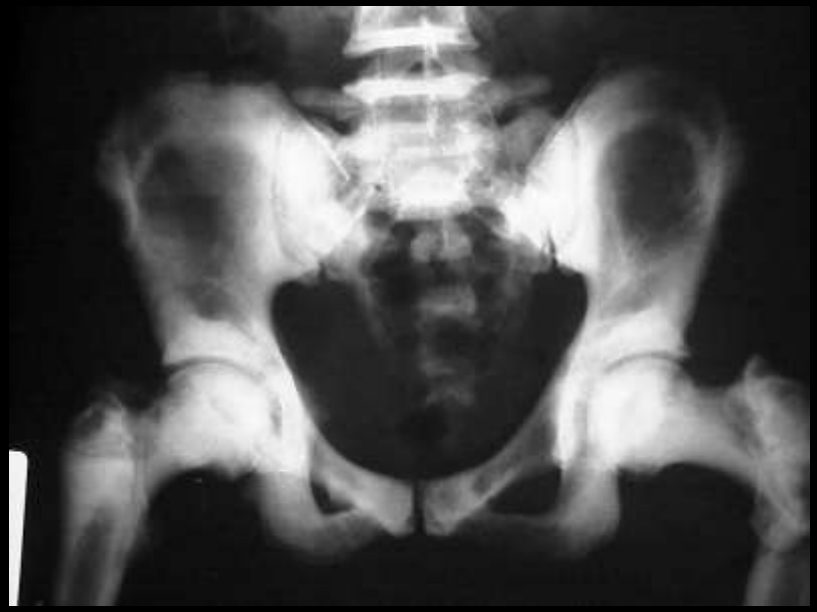


57M

Osteopetrosis

- Sclerotic, peri and endosteal
- Erlenmeyer flask
- Bone in bone, sandwich vertebrae
- Rugger jersey spine
- Calvaria and mandible spared
- Transverse metaphyseal bands
- Fractures



57M L hip pain

Osteopetrosis

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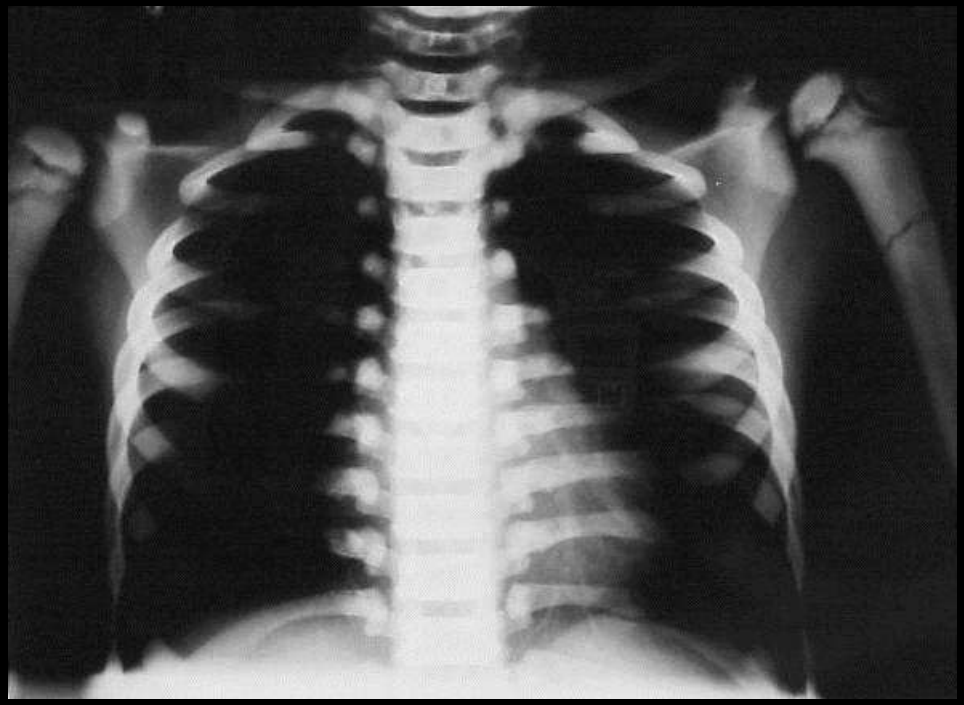


Bone in Bone

- Neonate – normal
- Growth arrest / recovery lines
- Paget's disease
- Osteopetrosis
- Acromegaly
- Heavy metal poisoning
- Prostaglandin E therapy
- AVN / Infarct
- Sequestrum

Erlenmeyer Flask Deformity

- Osteopetrosis
- Thalassemia / Sickle cell disease
- Gauchers / Niemann-Pick
- Metaphyseal dysplasia

Generalised Increase in Bone Density

- Dysplasias
  - Osteopetrosis
  - Pyknodysostosis
  - Craniotubular dysplasias (Pyle)
  - Craniotubular hyperostoses (Camurati-Engelmann)
- Metabolic
  - Renal Osteodystrophy
- Poisoning
  - Lead
  - Fluorosis
  - Hyper D
  - Hyper A
- Idiopathic
  - Caffey's disease
  - Idiopathic hypercalcaemia of infancy

Dysostosis Multiplex

- General name given to the bony appearances of the mucopolysaccharidoses and mucolipidoses

Dysostosis Multiplex

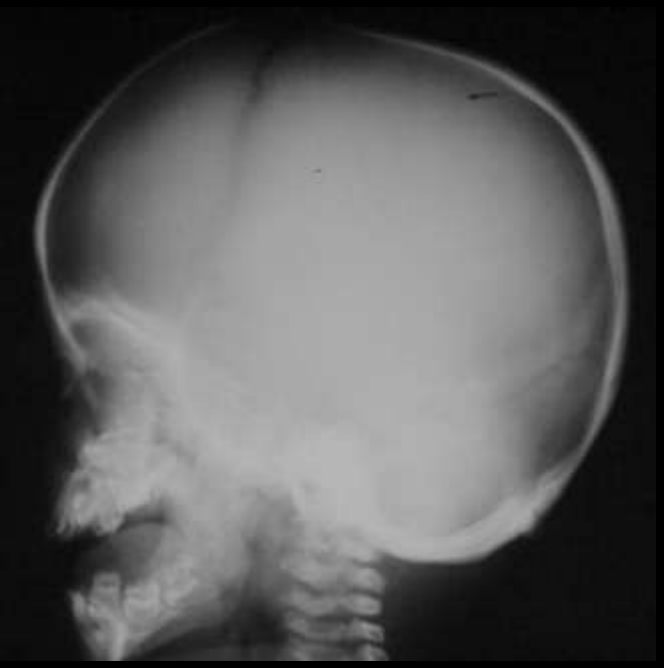
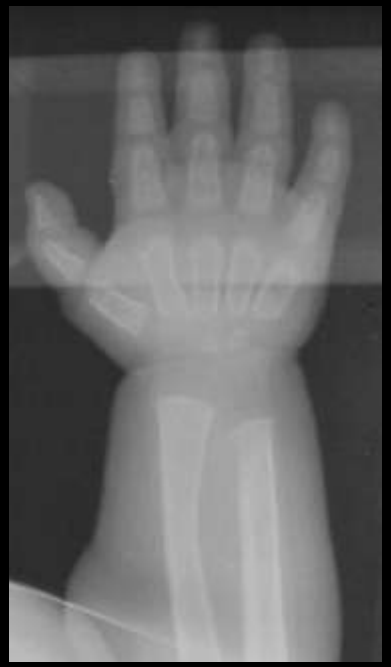
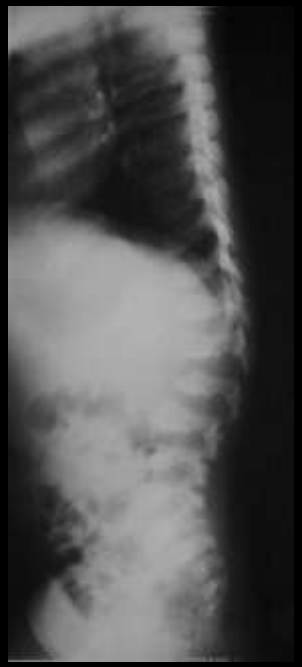
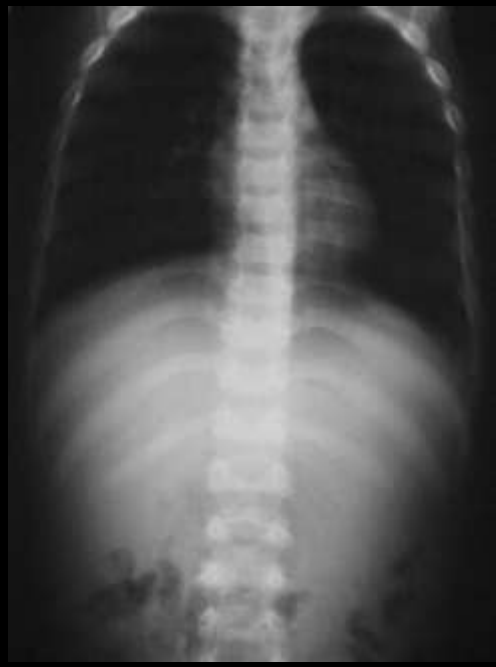
- Mucopolysaccharidoses
  - 1H – Hurler
  - 1S – Scheie
  - 2 – Hunter
  - 3 – Sanfilippo
  - 4 – Morquio
  - 5 – Maroteaux-Lamy
  - 6 – Sly
- Mucolipidoses
- Oligosaccharidoses

Dysostosis Multiplex

- Which one is different?

- Morquios
  - Not mentally retarded
  - Hypoplastic dens
  - Central anterior vertebral body beaks
  - Defective ossification of femoral heads with flattening

Dysostosis multiplex

Hunter Hurler syndrome 16mF

MPS 2 - Hunters Syndrome
General

- Similar to Hurlers syndrome, but:
- X linked recessive
- Later onset 2-6 years, death in 2nd or 3rd decade

MPS1H - Hurlers Syndrome
General

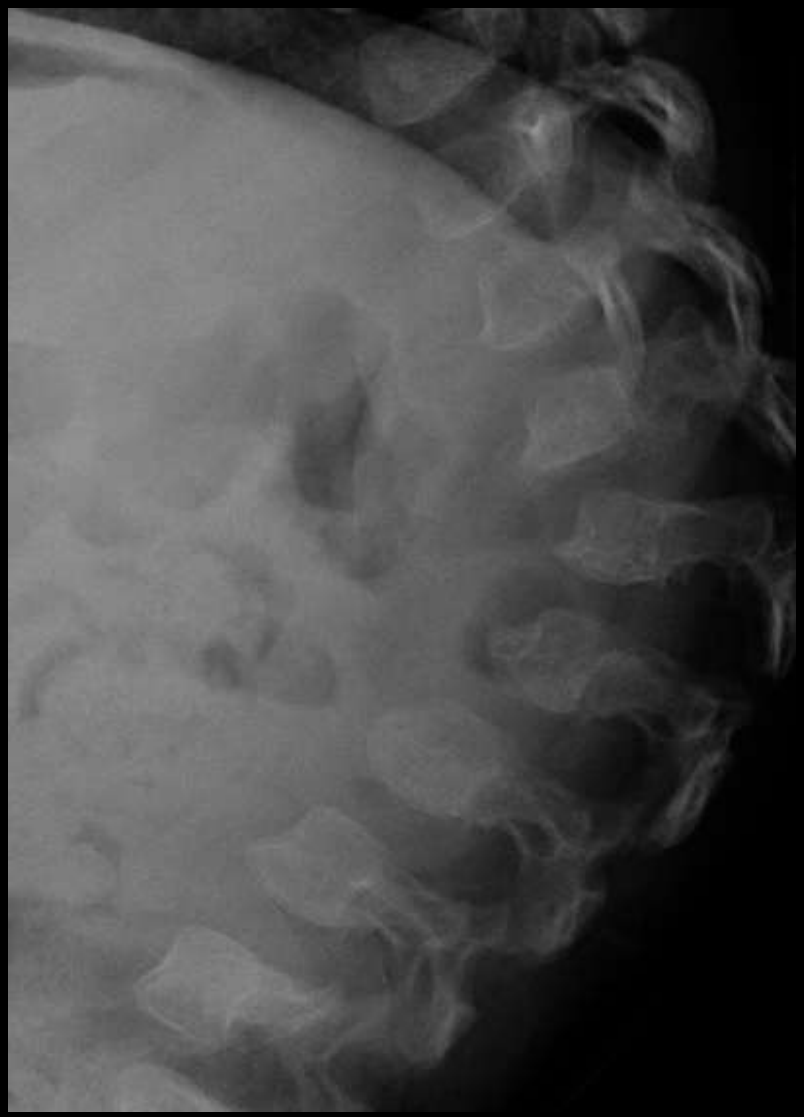
- MPS IH
- AR
- Dwarf, mental retardation,
- Death from cardiac failure in first decade

MPS1H - Hurlers Syndrome
Radiology - Skull

- Scaphocephalic macrocephaly
- J shaped sella

MPS1H - Hurlers Syndrome
Radiology – Axial skeleton

- Oval vertebral bodies with an anterior beak
- Kyphosis and a thoracolumbar gibbus
- Posterior scalloping with widened interpediculate distance
- Short neck



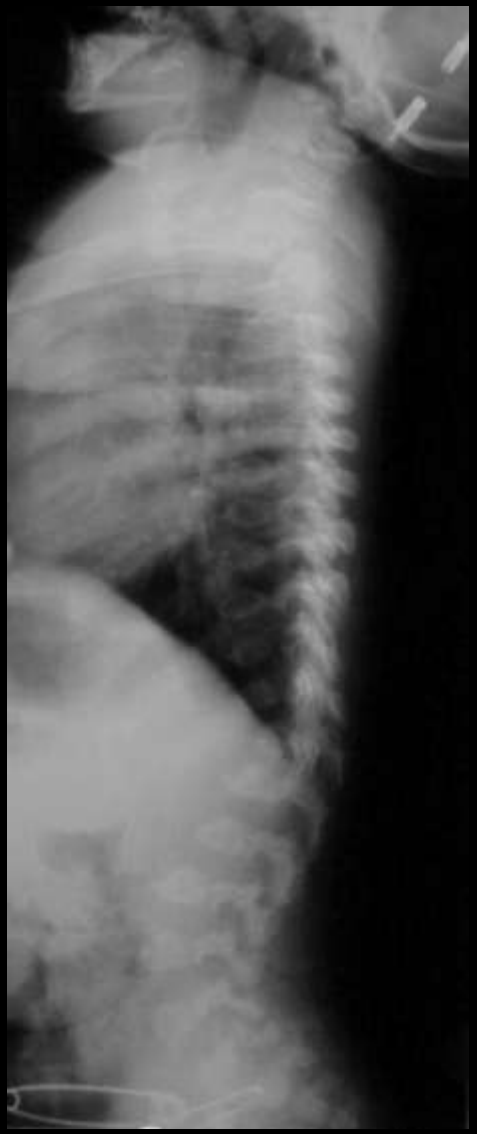
2 year old male w/ back pain.

MPS4 - Morquios Syndrome
General

- MPS IV
- AR
- Present 2nd year
- Decreassed growth
- Skeletal deformity

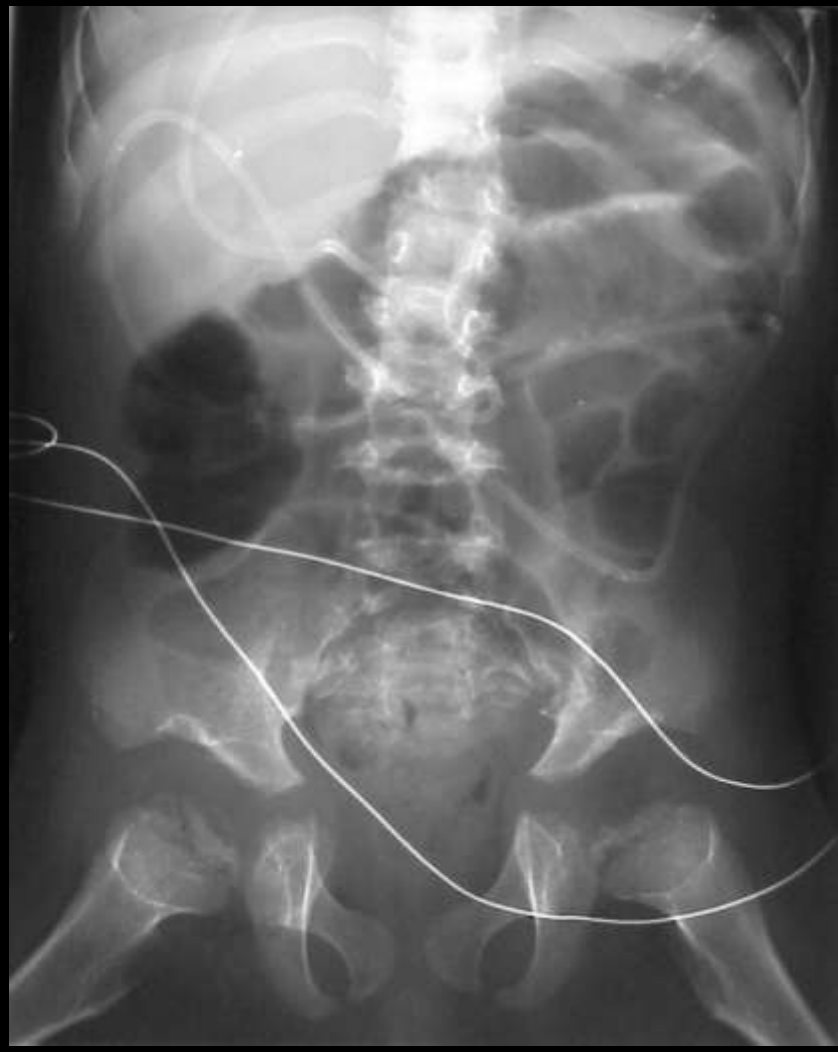
MPS4 - Morquios Syndrome
Radiology – Axial skeleton

- Universal vertebrae plana, wide discs
- Hypoplastic dens
- Hypoplastic thoracolumbar vertebrae displaced posteriorly
- Central anterior vertebral body beaks
- Short neck
- Thoracic scoliosis and thoracolumbar kyphosis



MPS4 - Morquios Syndrome
Radiology – Appendicular skeleton

- Defective ossification of femoral heads with flattening
- Genu valgum
- Short wide tubular bones with irregular metaphyses
- Proximal tapering of metacarpals
- Irregular carpal and tarsal bones



Causes of bilateral femoral head collapse/fragmentation children

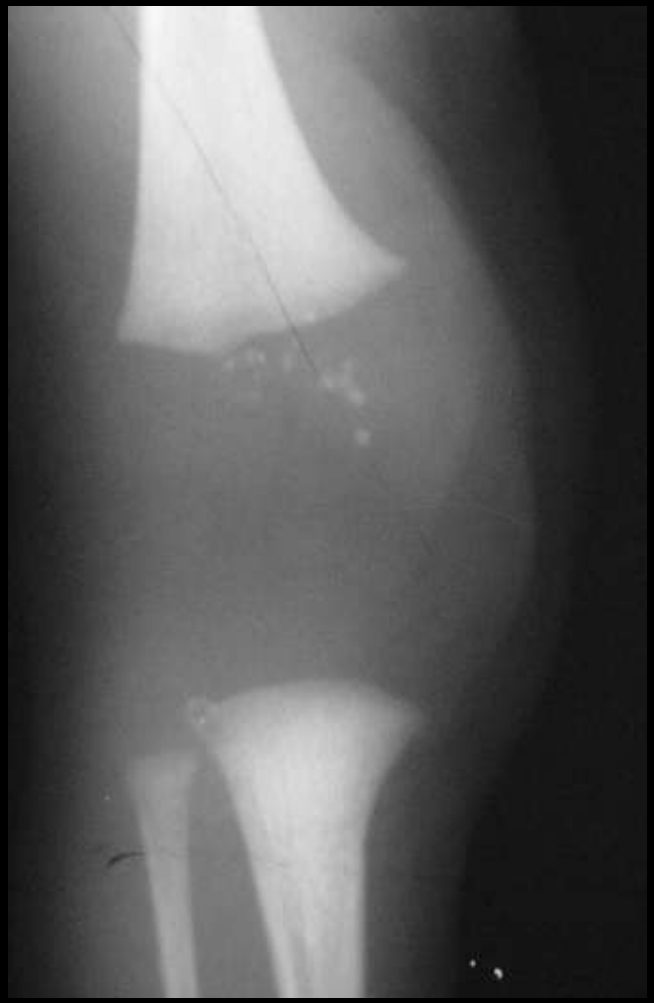
- AVN Perthes 10% bilateral
- Multiple epiphyseal dysplasia
  - Myers dysplasia (just femoral heads)
- Gauchers
- Morquios
- Hypothyroidism (Cretinism)
- Chondrodysplasia punctata

Chondrodysplasia Calcificans Punctata

- AKA
  - Chondrodysplasia punctata
  - Conradi- Hunerman
- AD or AR, Presents – Birth, early infancy
- Clinical
  - Short limbs - asymmetrical
  - Flat face
  - Joint contractures
  - Ichthyosiform skin
  - Congenital heart disease

Chondrodysplasia Calcificans Punctata Autosomal Dominant – Conradi Hunnerman

- Stippled epiphyses
  - Ends of long bones
  - Carpal and Tarsal
- Later develop into epiphyseal dysplasia
- Unilateral shortening of tubular bones
- Coronal cleft in vertebral bodies



2dF

Chondrodysplasia Calcificans Punctata Autosomal Dominant - Conradi

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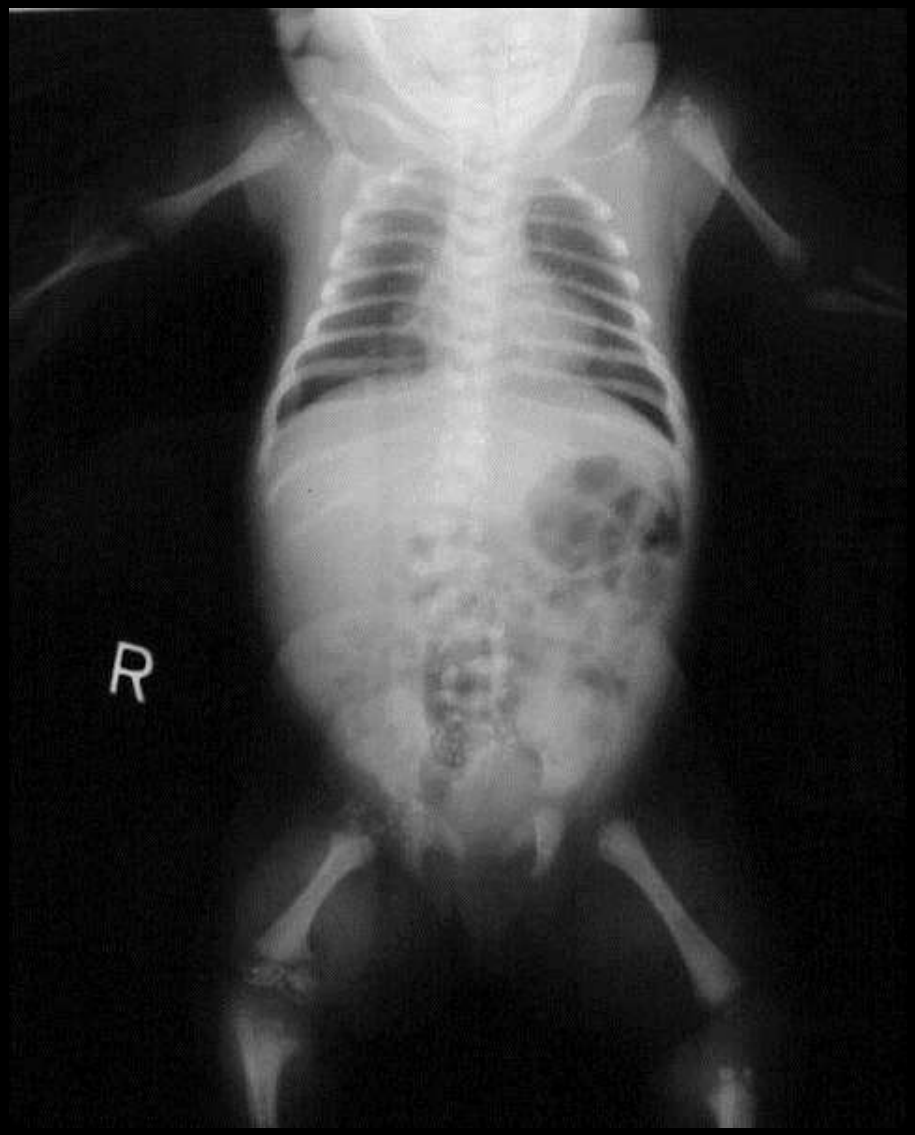

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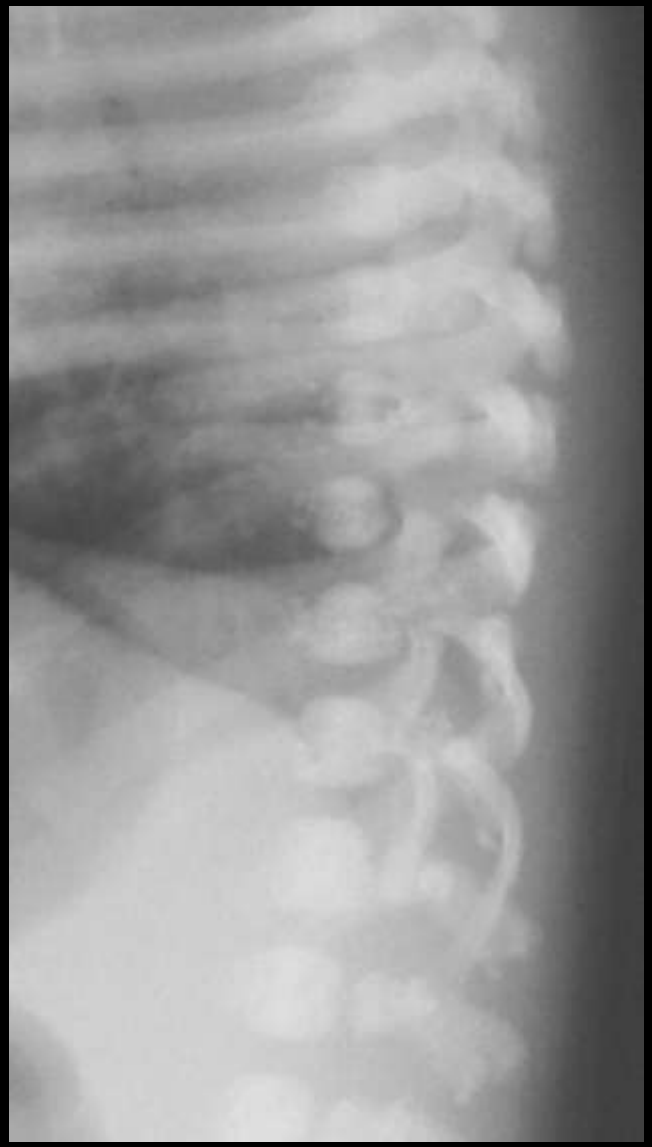

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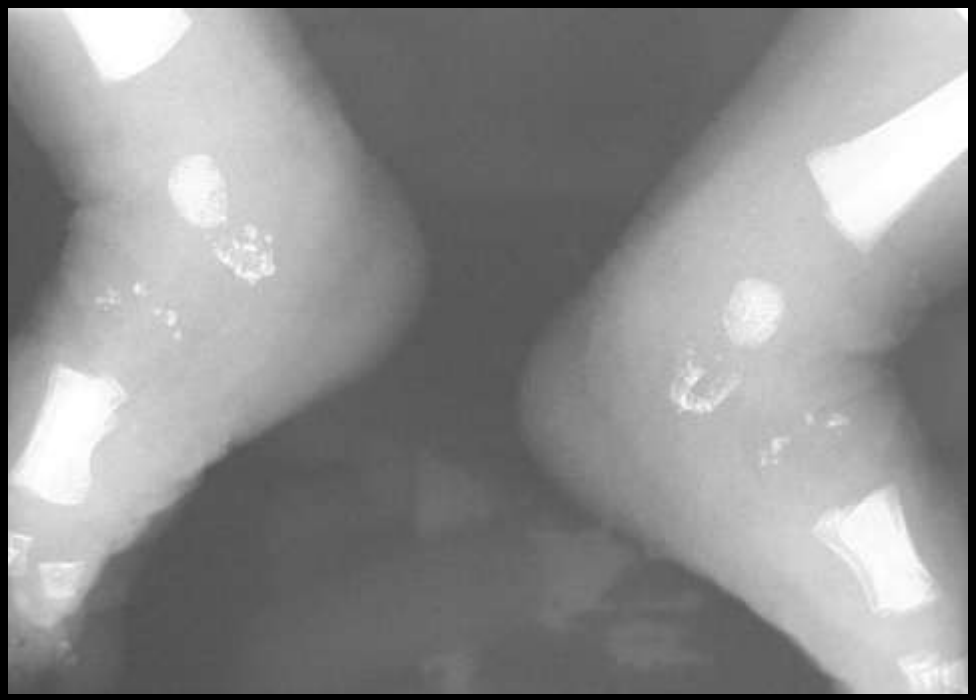

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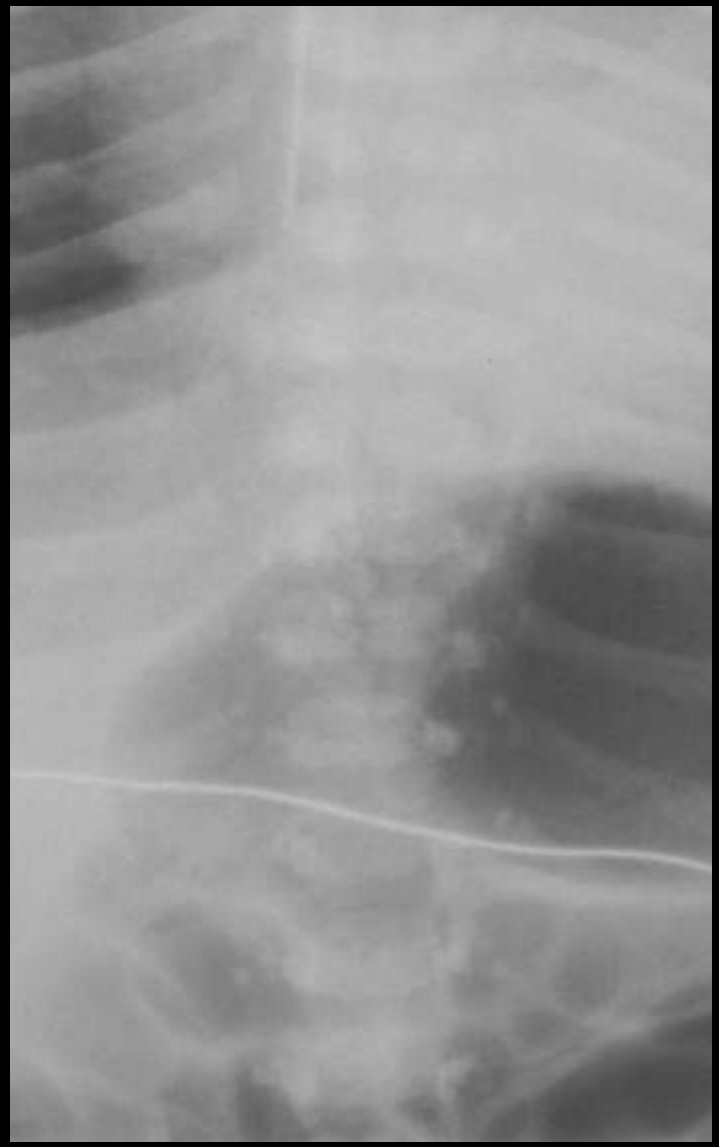
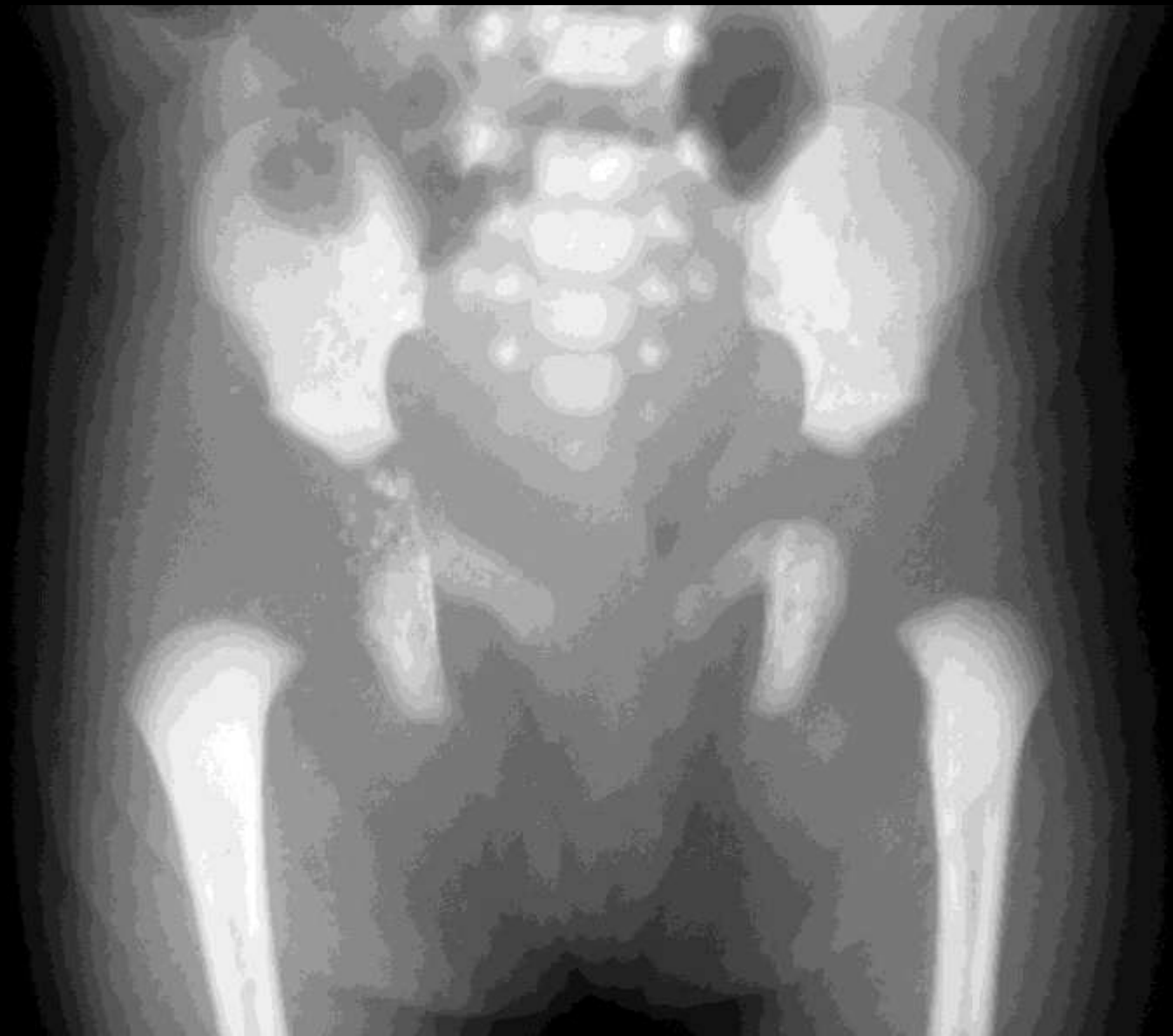
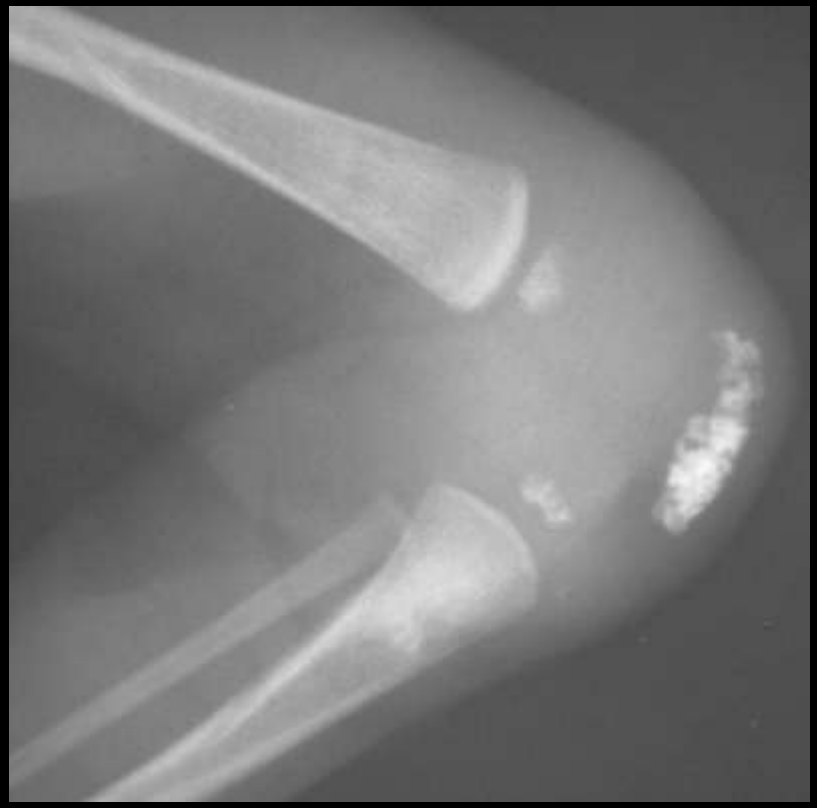


Chondrodysplasia Calcificans Punctata Autosomal Dominant - Conradi

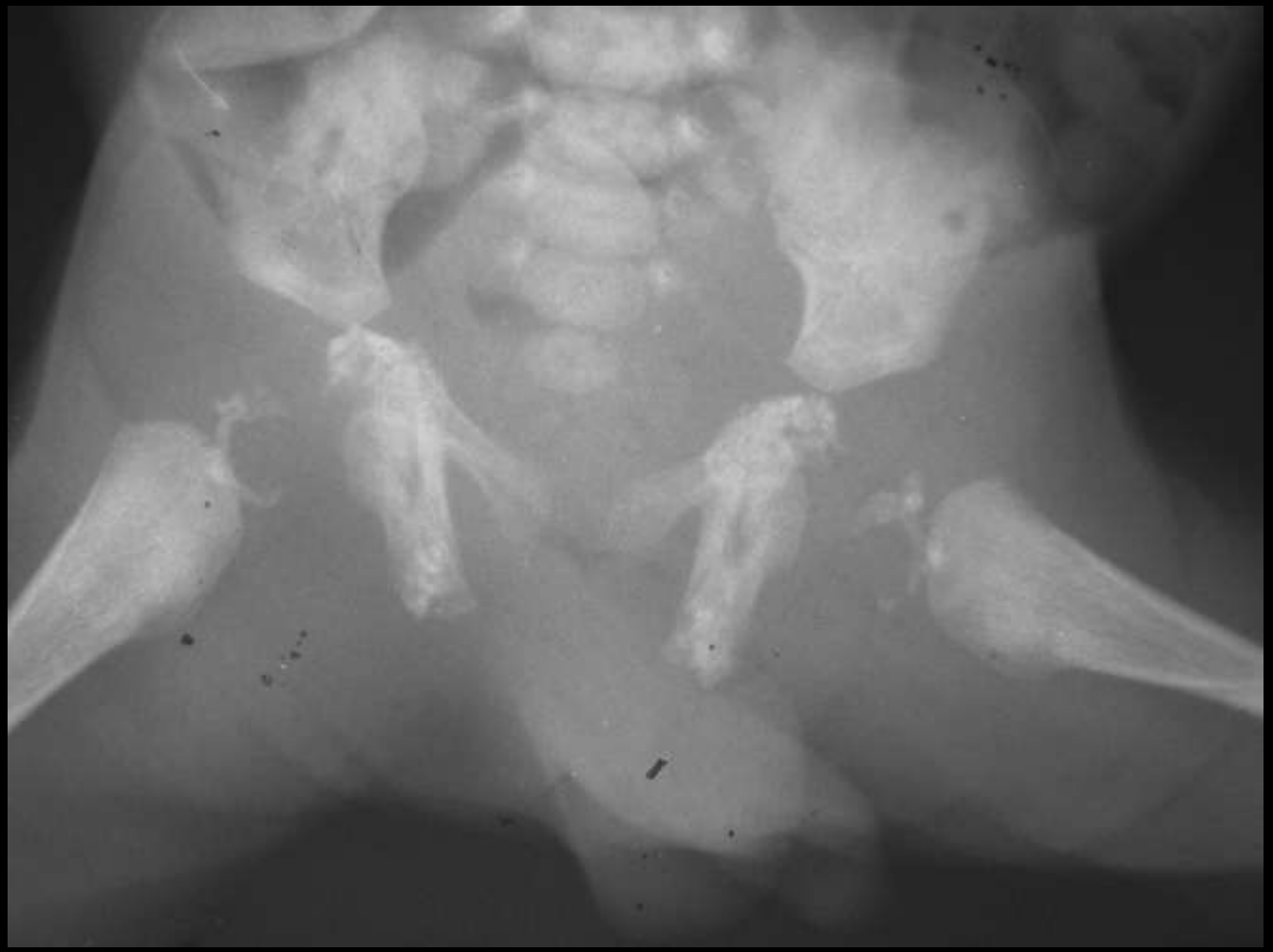


Chondrodysplasia Calcificans Punctata Autosomal Recessive

- Severe symmetric shortening
- Metaphyseal splaying
- Calcific stippling
- Dorsal and ventral ossification centers of vertebrae seperate



Chondrodysplasia Calcificans Punctata Autosomal Recessive

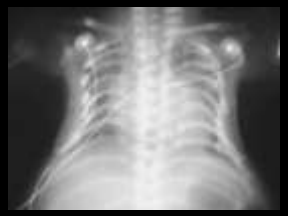


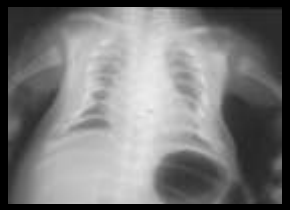
Causes of Stippled Epiphyses

- Normal
  - Distal femur
- AVN Perthes 10% bilateral
- Multiple epiphyseal dysplasia
  - Myers dysplasia (just femoral heads)
- Morquios – causes flattening
- Hypothyroidism (Cretinism) delayed onset
- Chondrodysplasia punctata
- Trisomy 18 and 21
- Warfarin embryopathy – disappears after 1 year

Thanatophoric Dwarfism

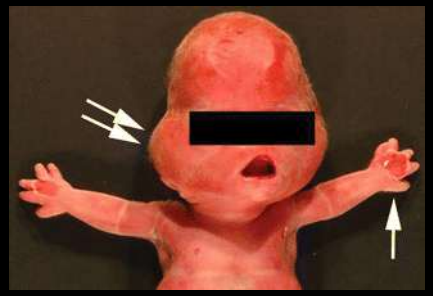
- Small thorax





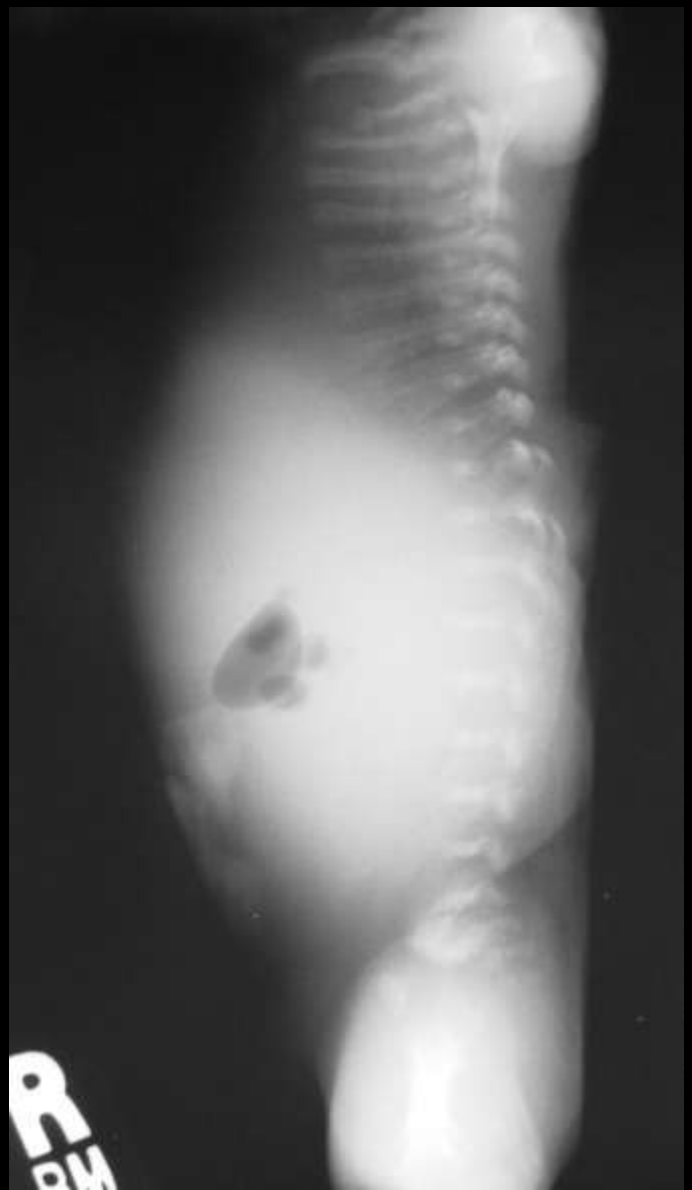
- Severe platyspondyly
  - H or inverted U shaped vertebrae
- Telephone handle shaped long bones

- Cloverleaf skull
  - Kleeblattschadel



Thanatophoric Dwarfism

- Small thorax
- Severe platyspondyly
  - H or inverted U shaped vertebrae
- Telephone handle shaped long bones
- Cloverleaf skull
  - Kleeblattschadel



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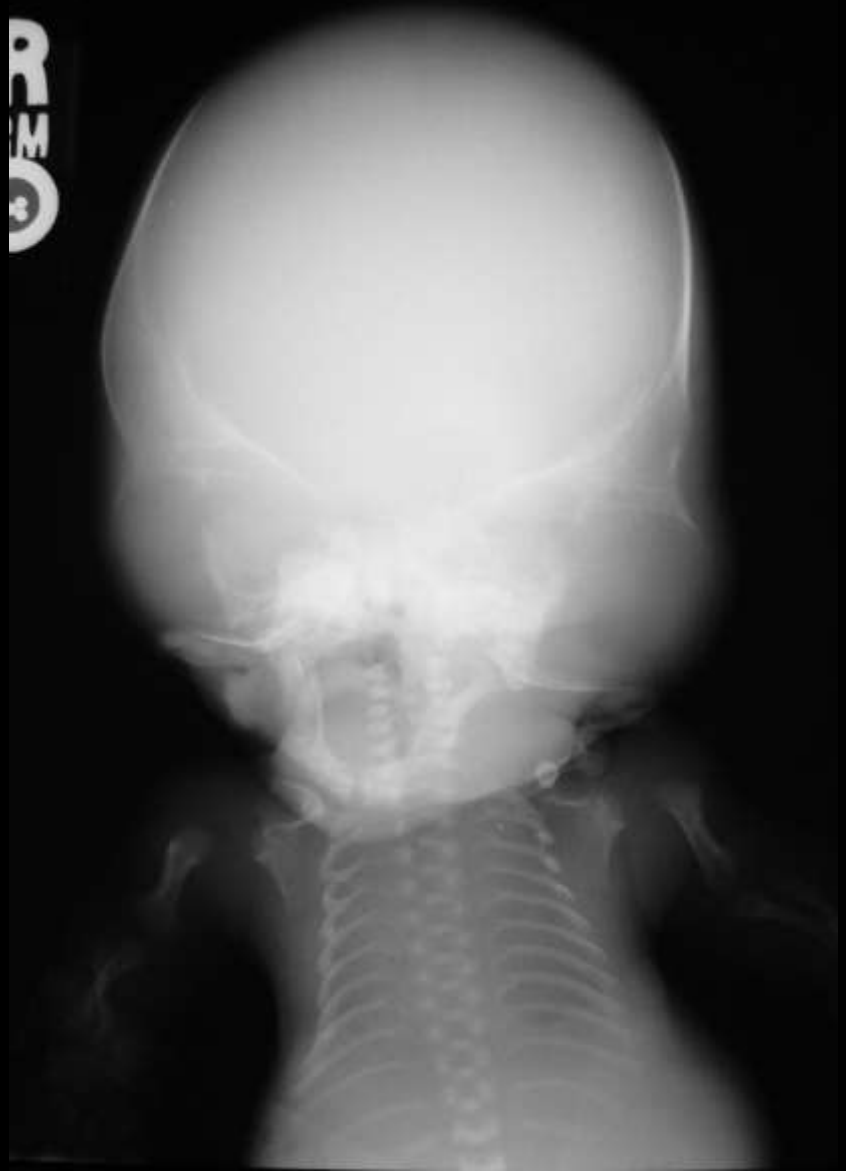

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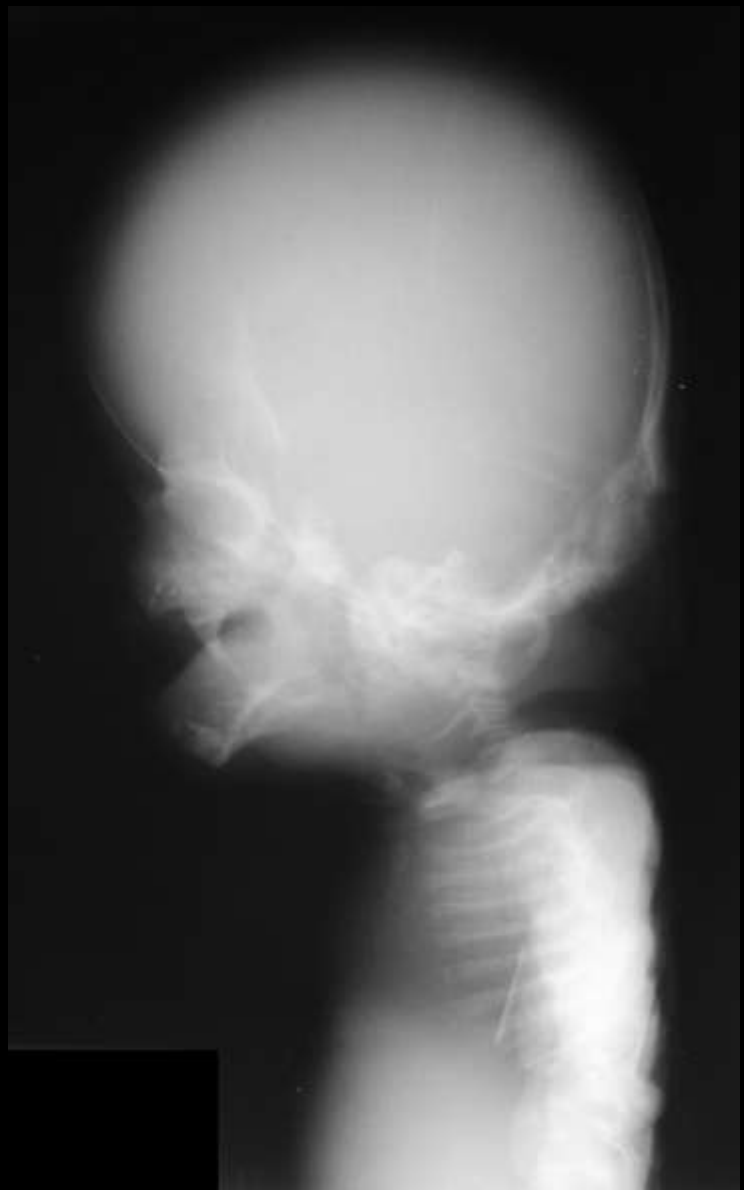

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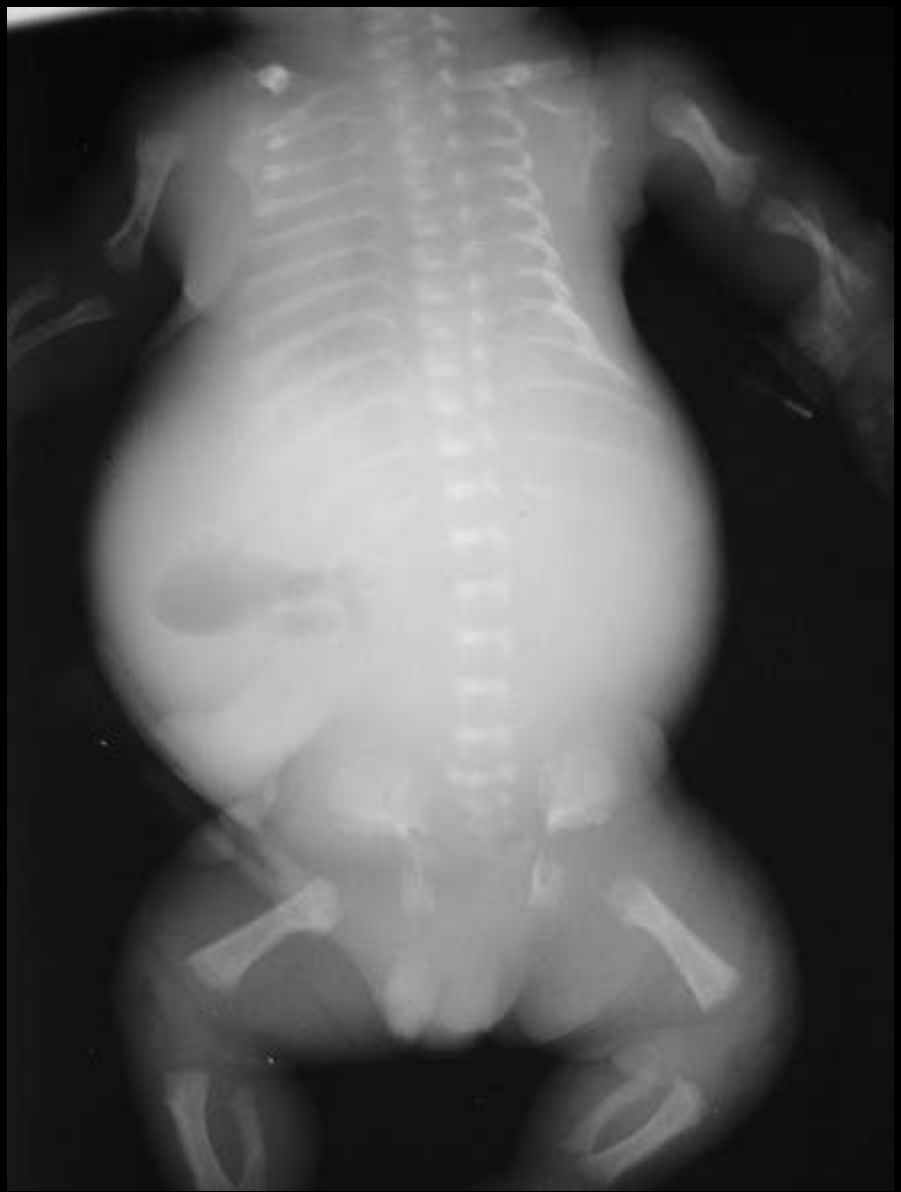
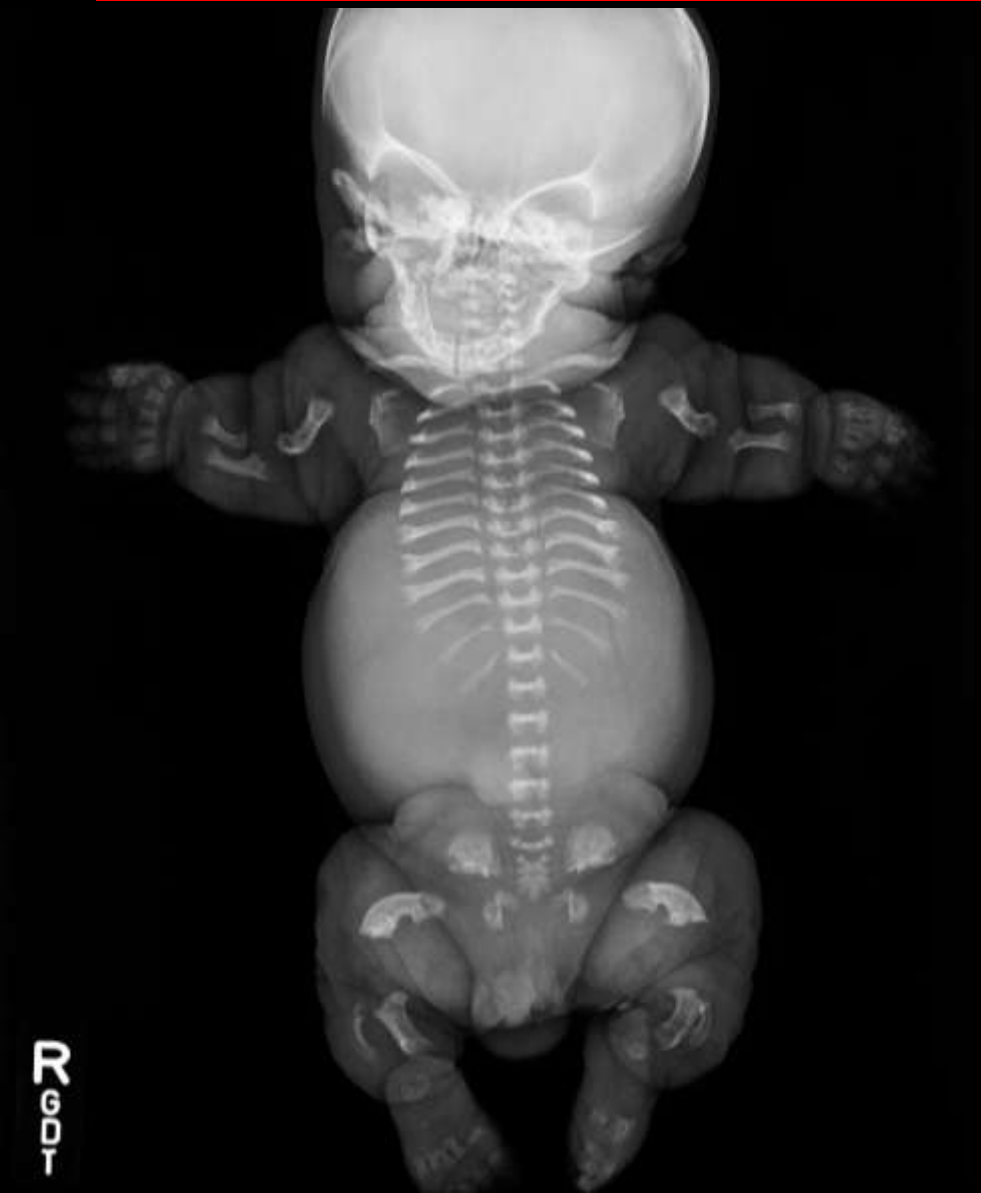


Lethal Neonatal dysplasias

- Osteogenesis imperfecta type 2
- Thanatophoric dwarfism
- Chondrodysplasia punctata AR
- Asphyxiating thoracic dystrophy
  - Jeune's syndrome
- Campomelic dwarfism
- Achondrogenesis
  - Homozygous achondroplasia
- Hypophosphatasia

Thanatophoric dysplasia



Chest

- Narrow; short horizontal ribs
- Cupped anterior ends
- Small scapulae with normal clavicles

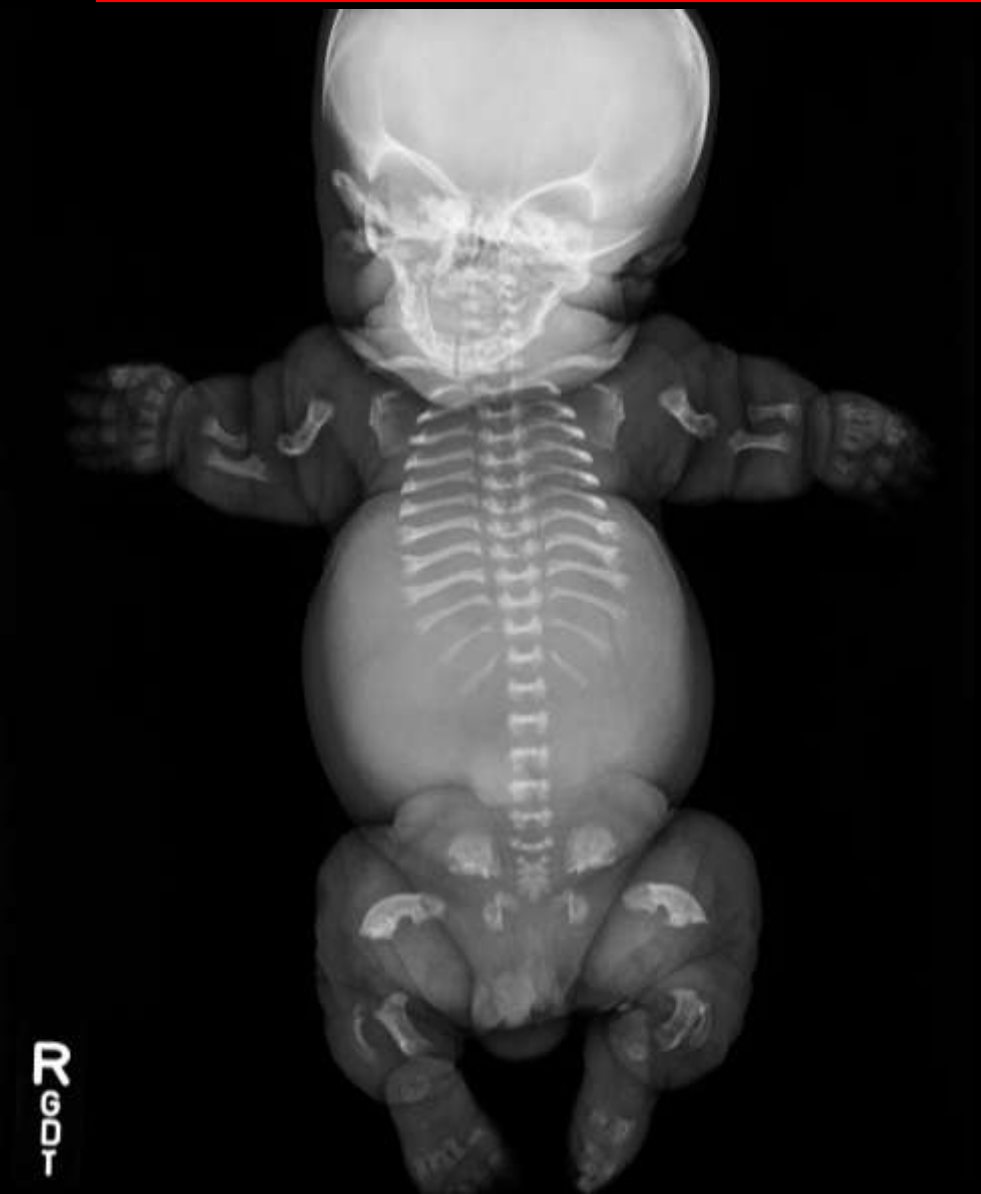
Pelvis

- Small, square iliac wings
- Flat acetabulae

Extremities

- Bowing of extremities
- Metaphyseal flaring
  - “telephone receiver”

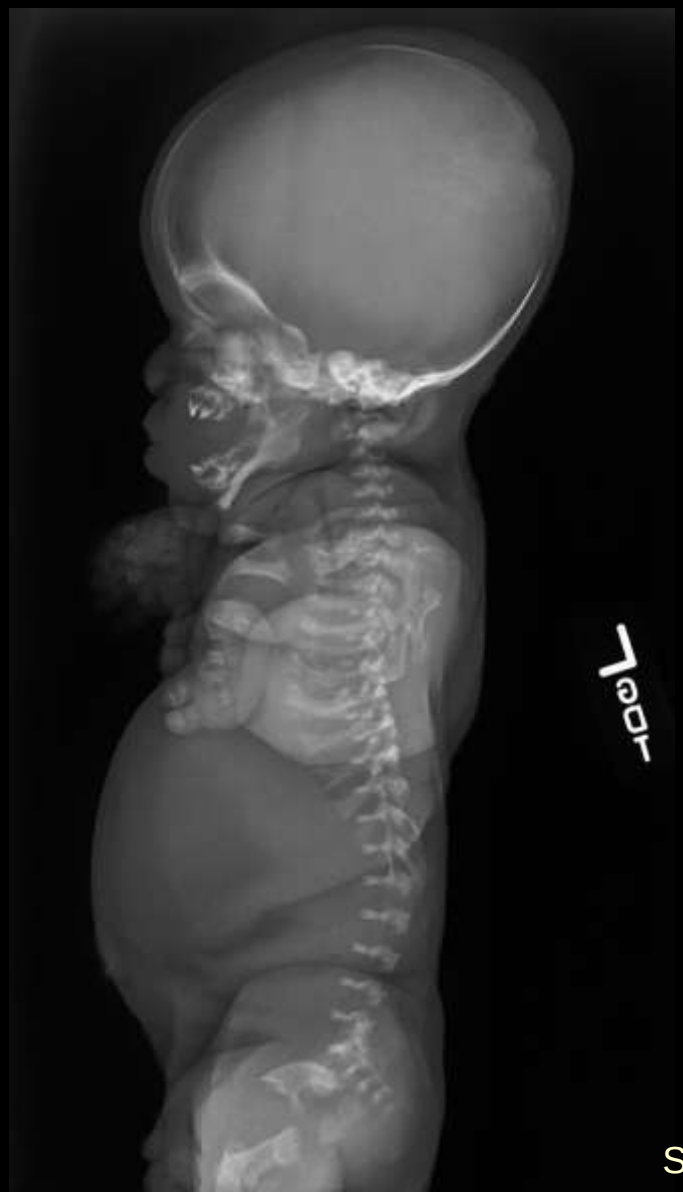
Thanatophoric dysplasia



- Spine
  - Flattened/H-shaped vertebral bodies
  - Normal trunk length

Thanatophoric dysplasia

- Macrocrania
- Flattened nasal bridge
- Frontal bossing
- Protruberant abdomen
- Platyspondyly



Stillbirth

Diastrophic Dysplasia

1. First described in 1960 by Lamy and Maroteaux
2. Prevalence – rare, but 1 in 30K in Finland
3. Autosomal recessive
4. Clinical Findings
    1. Cystic ear swelling
    2. Hitchhiker thumb
    3. Prominent cheeks
    4. Cleft palate

Diastrophic Dwarfism (Dysplasia)

- Clinical
  - AR, presents at birth
  - Small stature
  - Short extremities
  - Clubfoot
  - Hitchhiker’s thumb
  - Joint contractures

Diastrophic Dwarfism (Dysplasia)

- Short clubbed long bones
- Epiphyses may be flat and stippled
- Severe talipes equinovarus
- Short thick metatarsals
- Short 1st metacarpal
- Scoliosis
- Occ posterior scalloping

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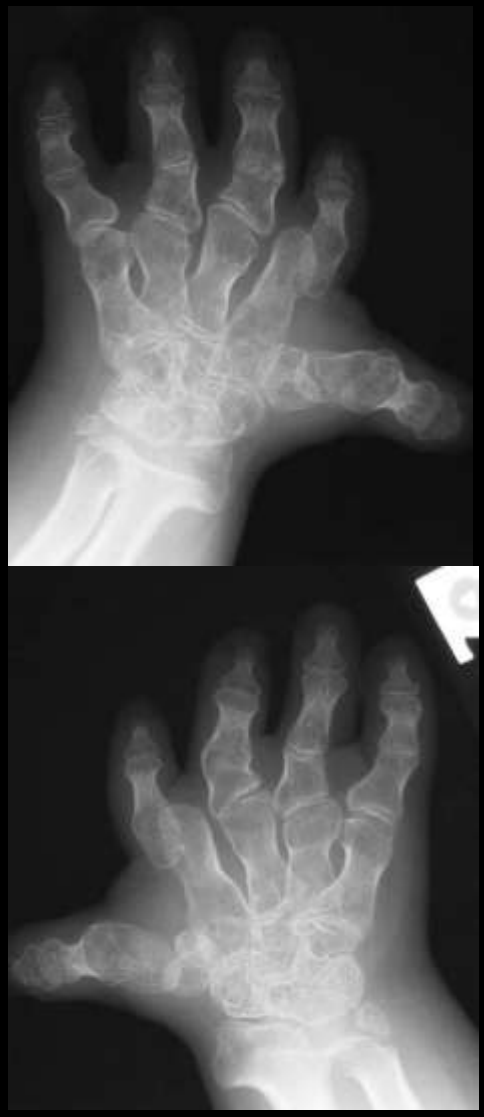

29F Short stature

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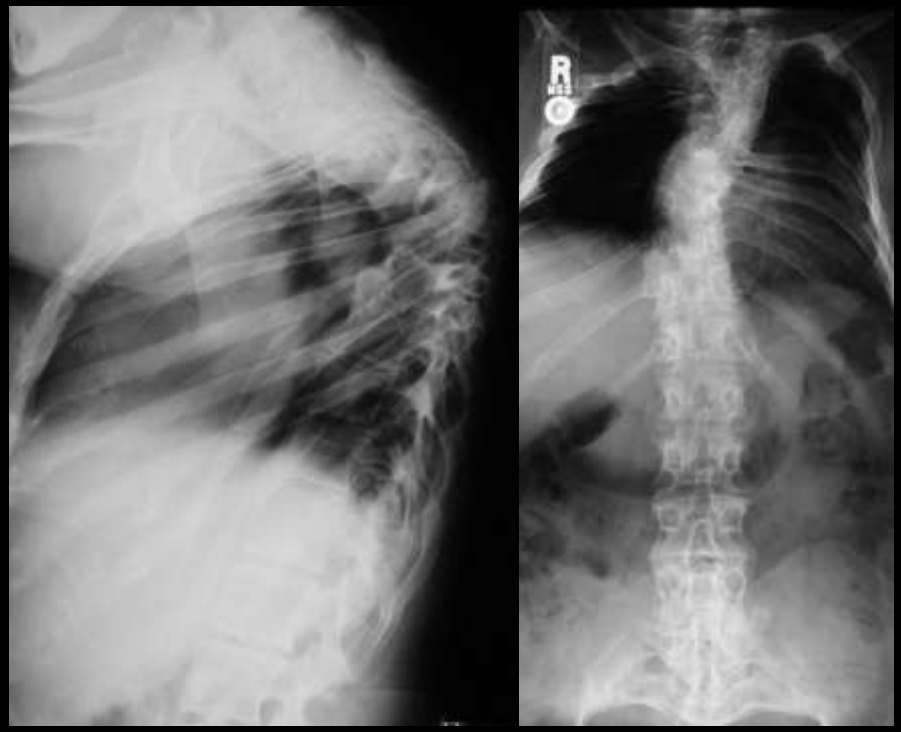

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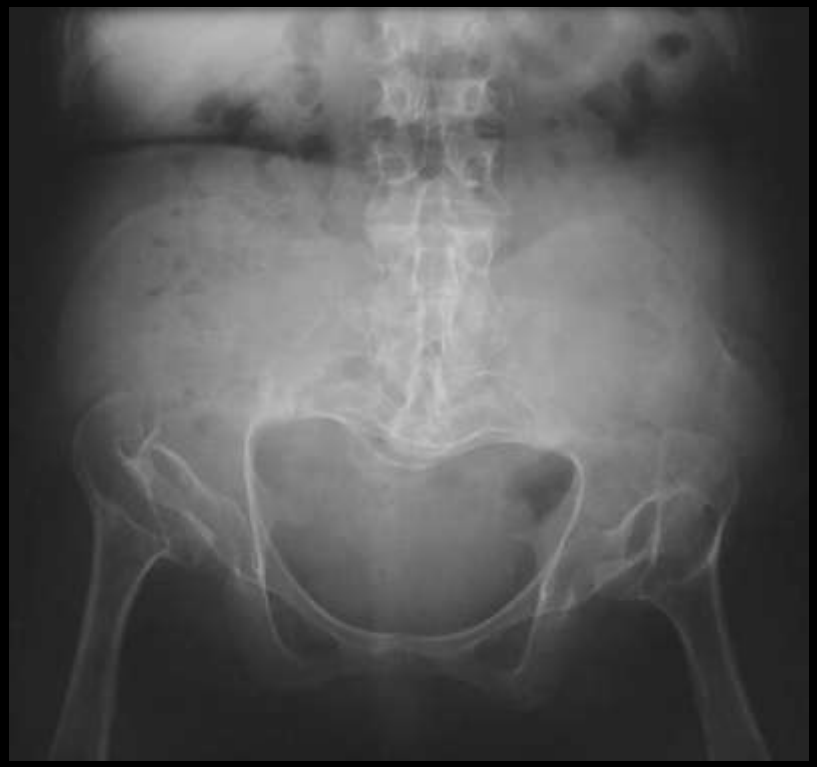

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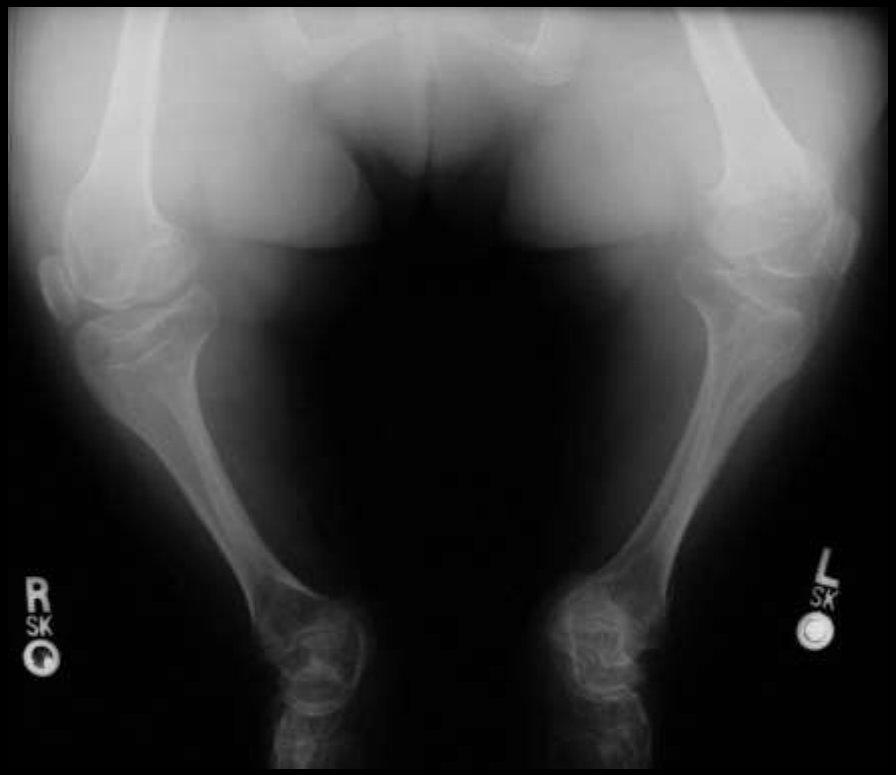

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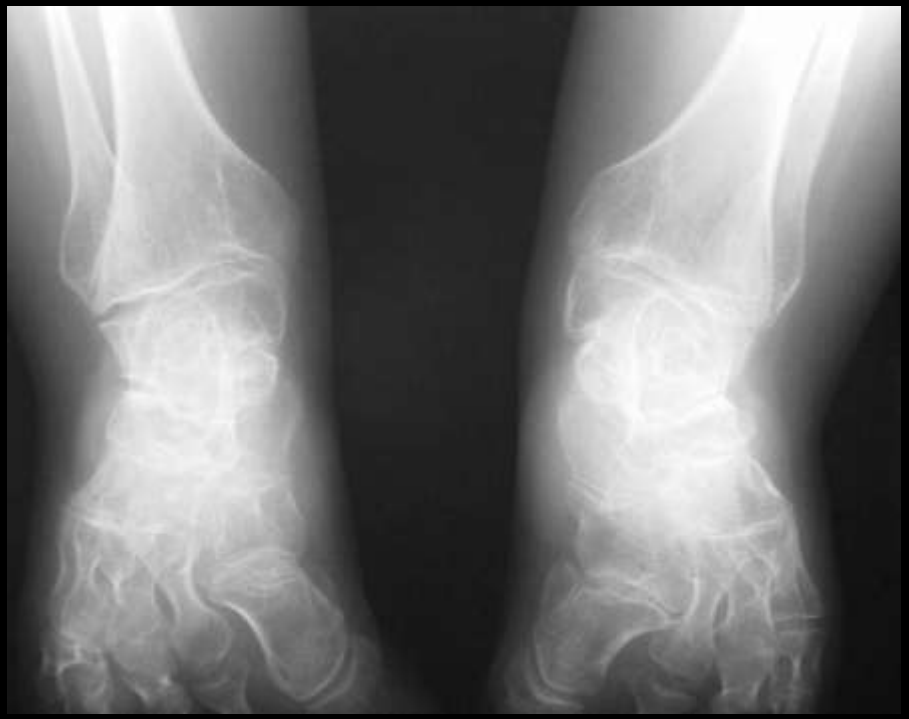

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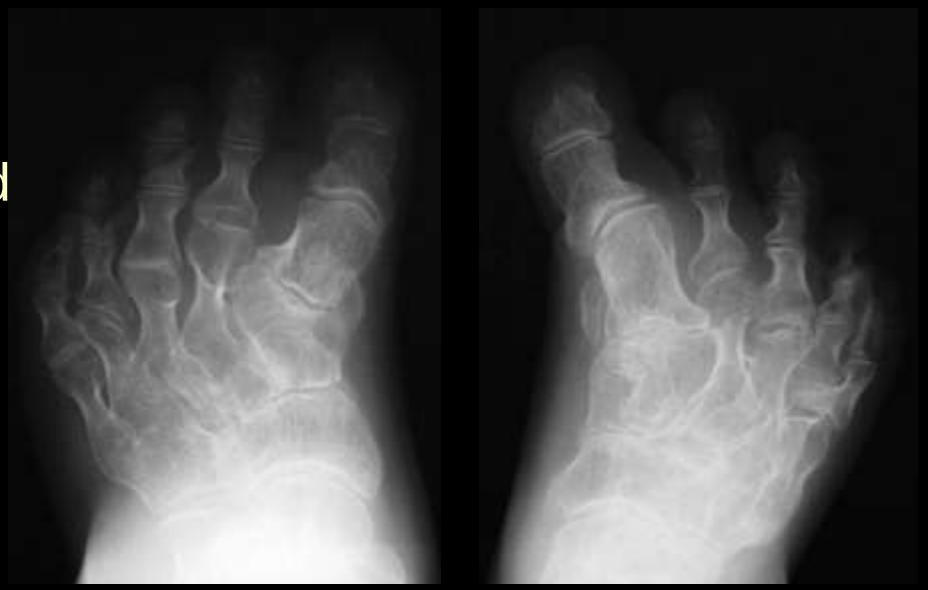

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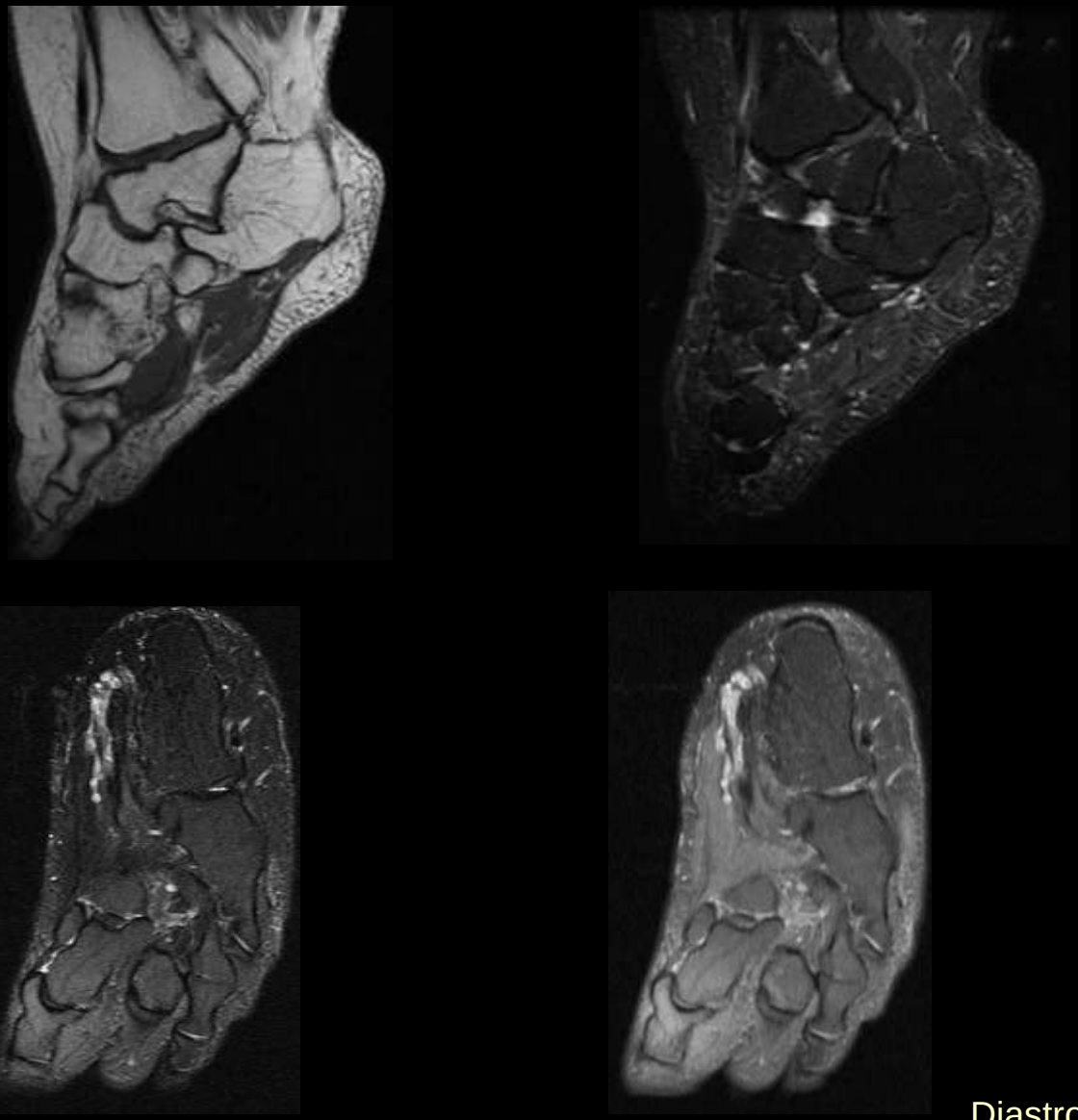
Diastrophic Dwarfism (Dysplasia)

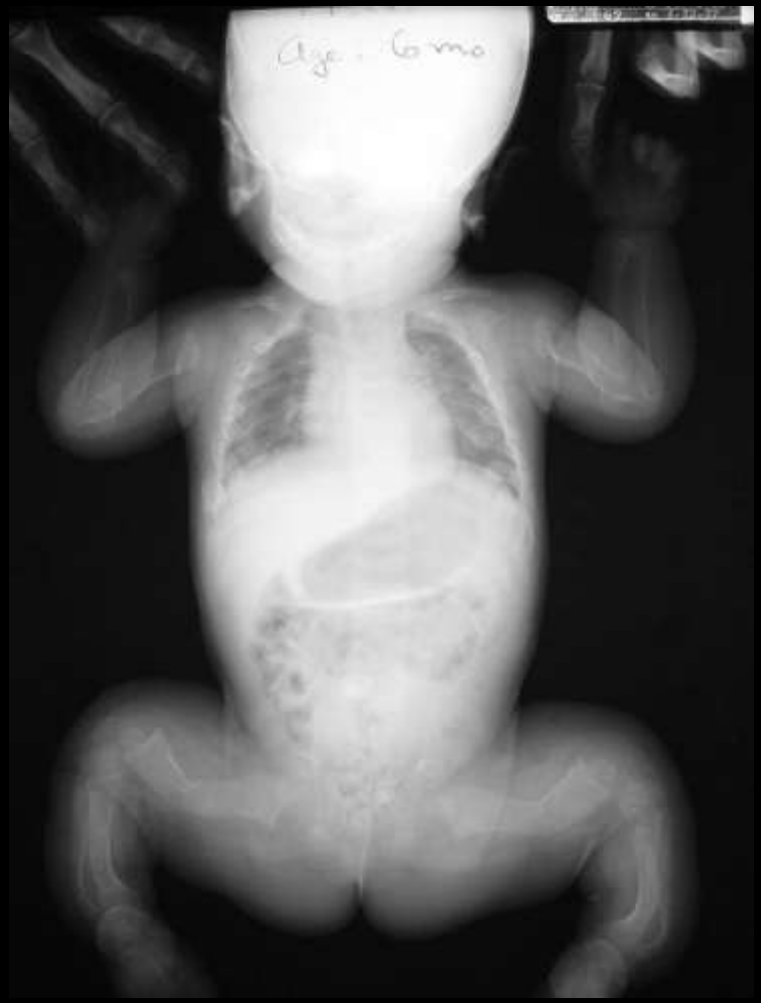


Diastrophic dwarfism 34F

OGI

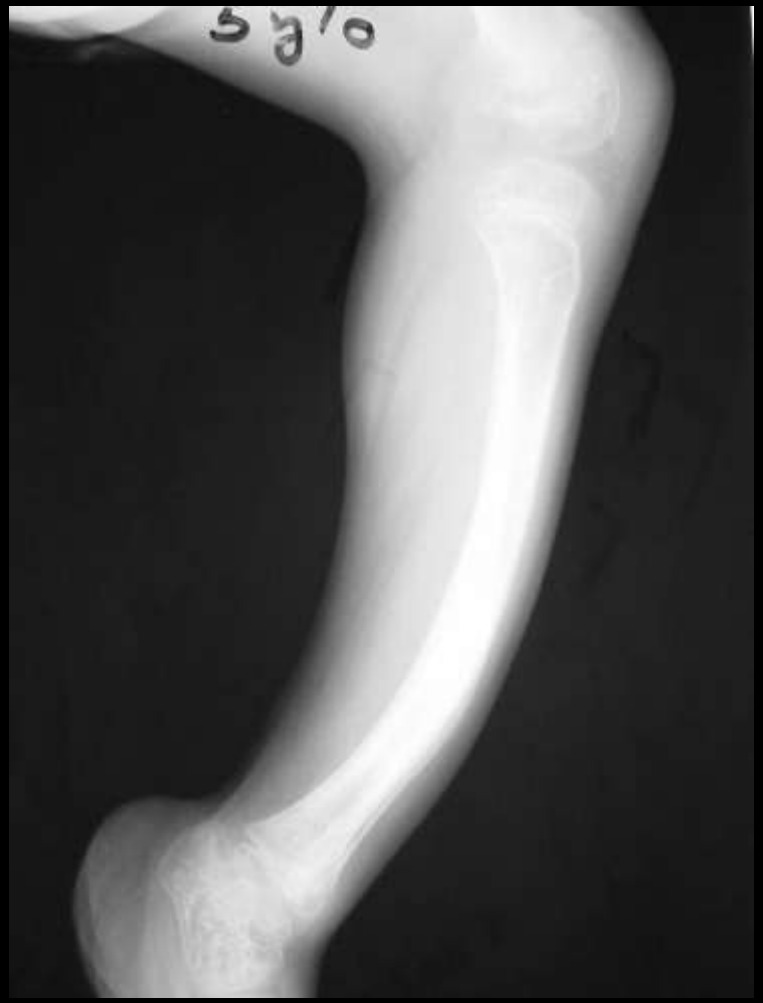
- Collagen disorder
- Fibrogenesis imperfecta
- Osseous fragility with fractures
- Rapid healing with much callus
- Blue sclera
- Wormian bones
- Osteopenia of skull
- Broad beaded ribs

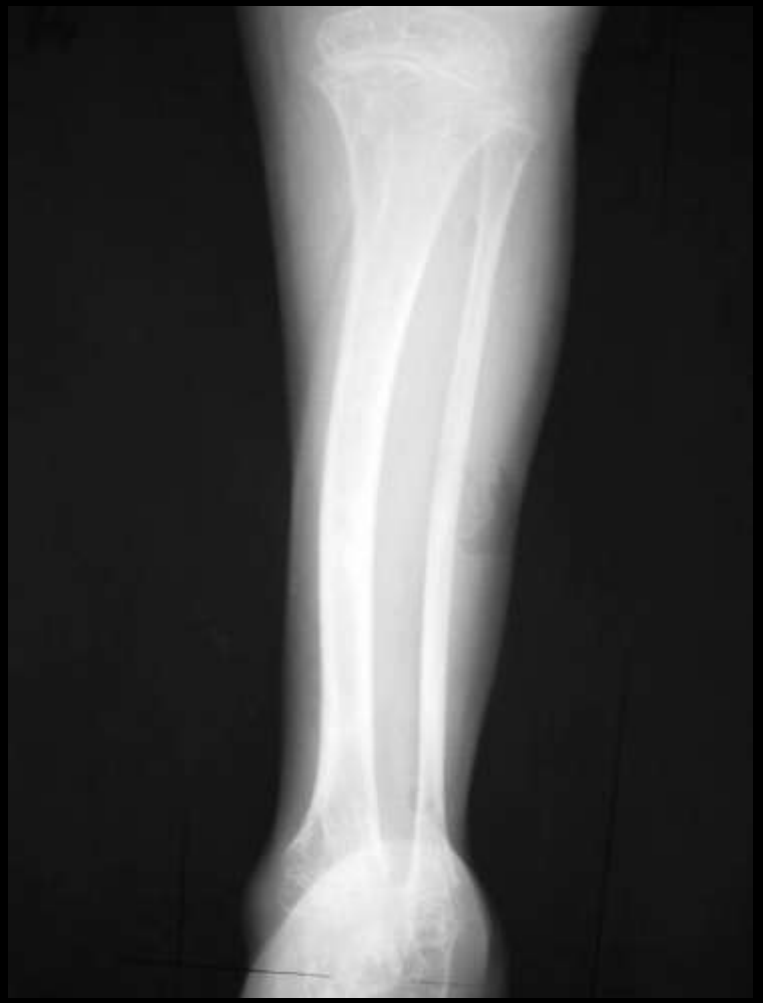
Osteogenesis Imperfecta



6mF

Osteogenesis Imperfecta





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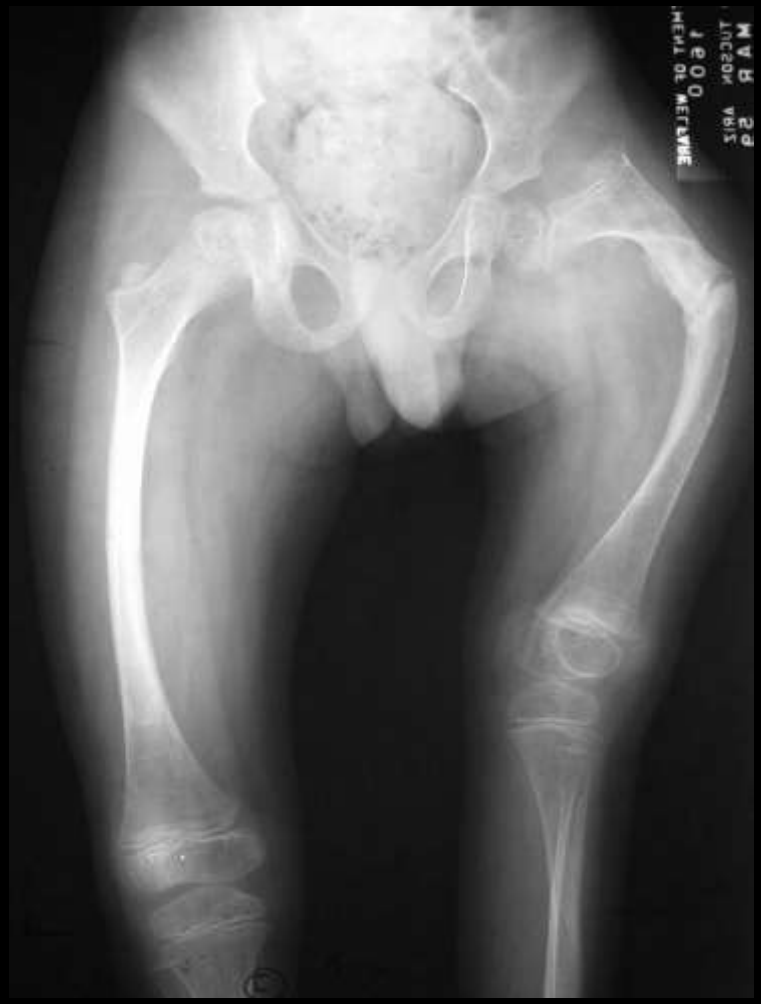

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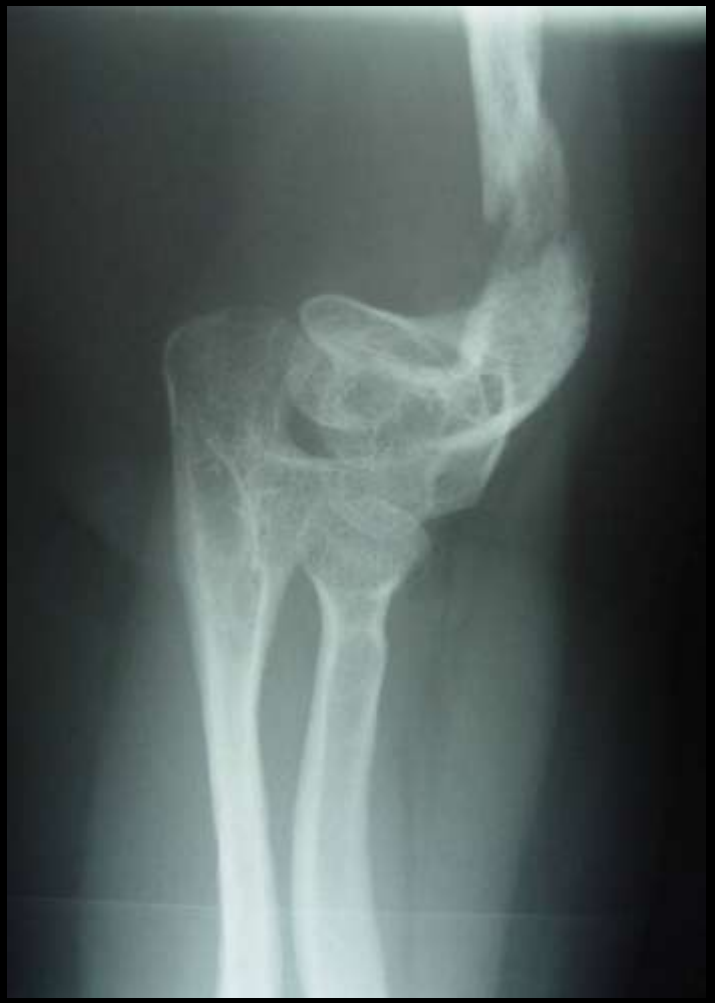
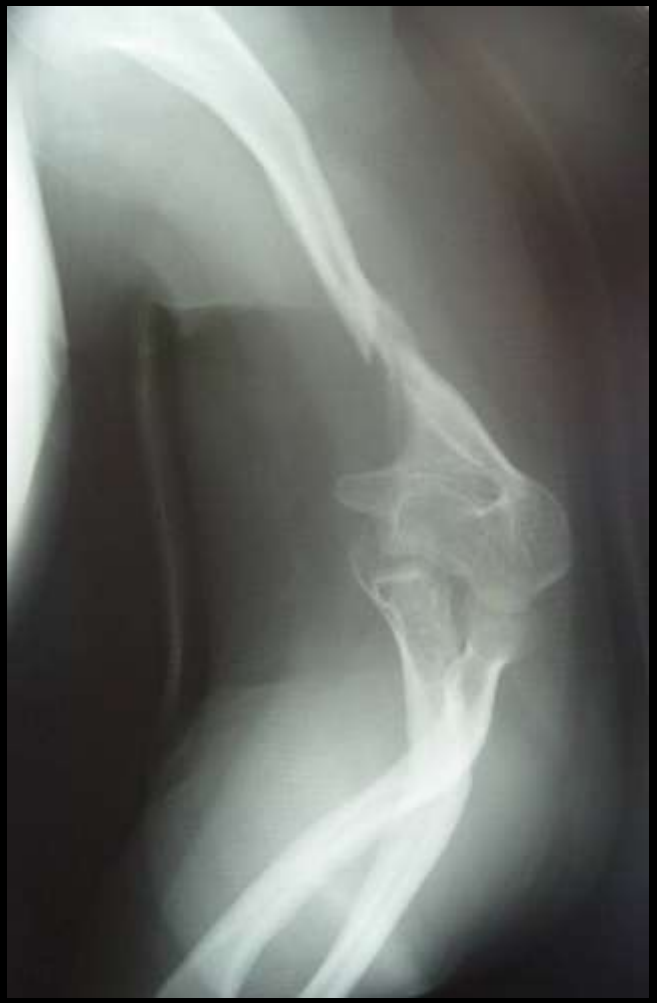
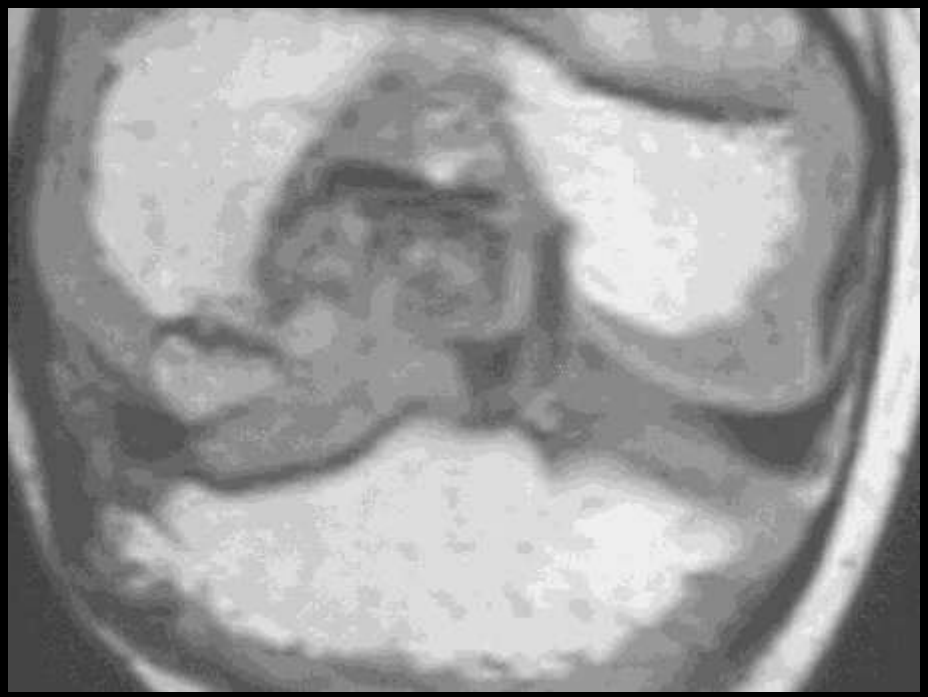
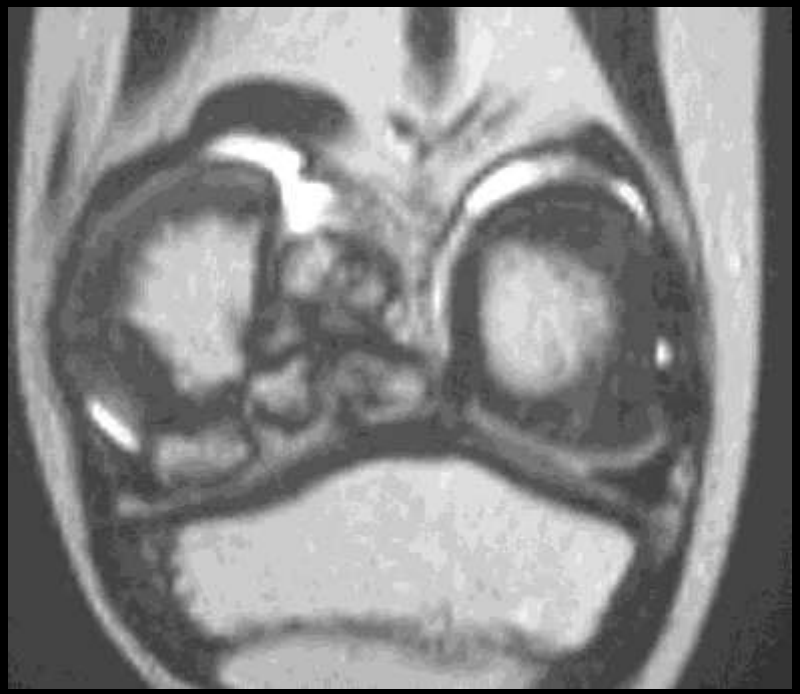




Dysplasia Epiphysialis Hemimelica
Trevor's Disease

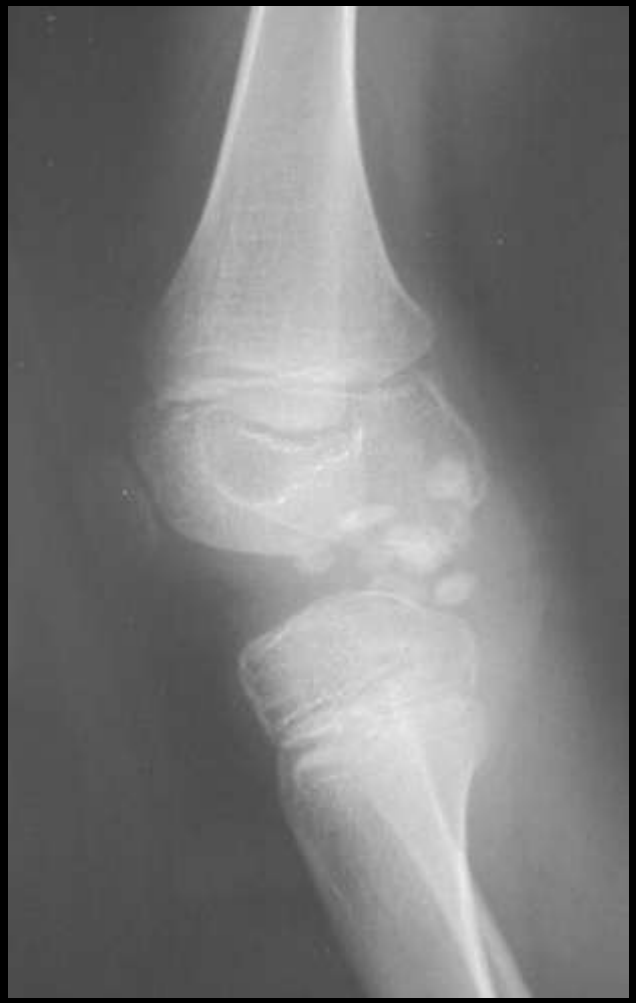


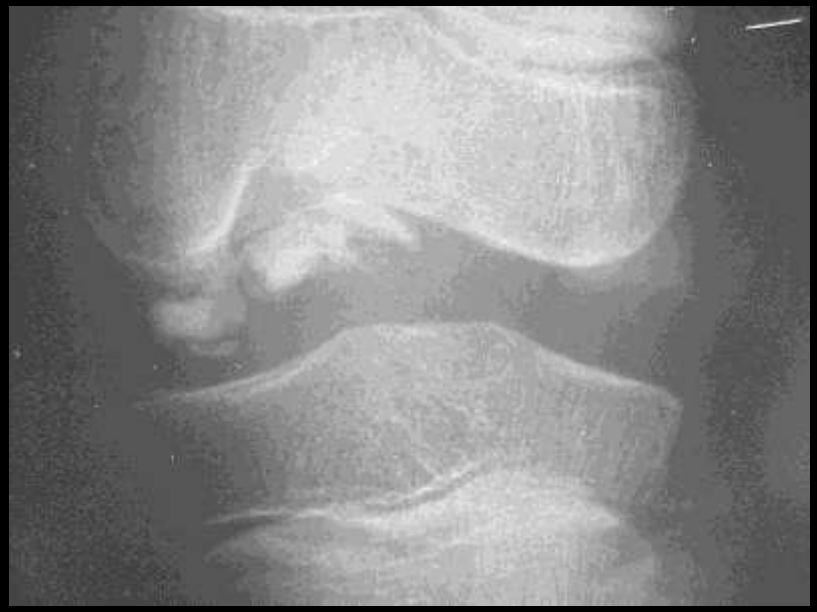
Cor T1



Cor T2

Dysplasia Epiphysialis Hemimelica
Trevor's Disease





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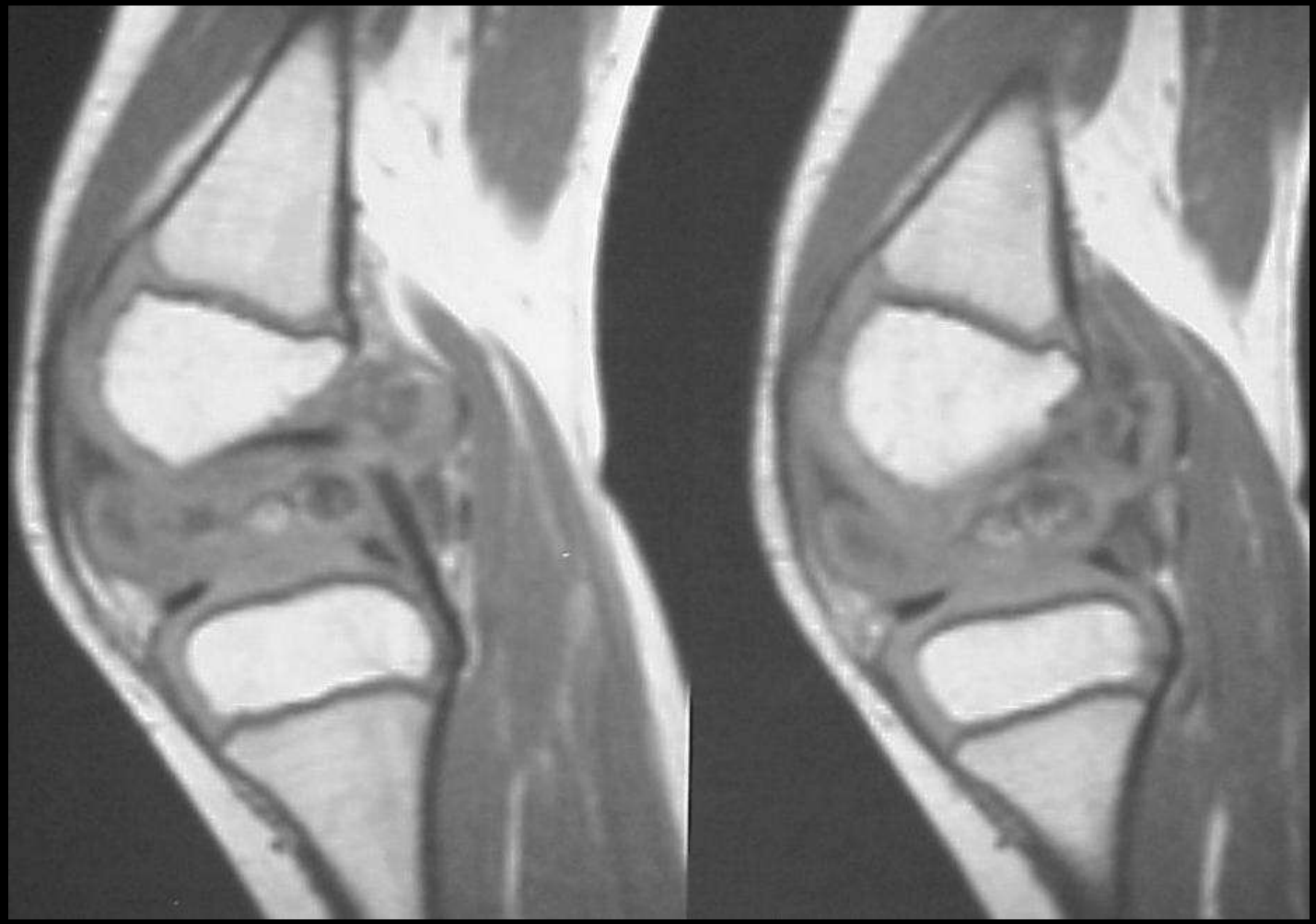
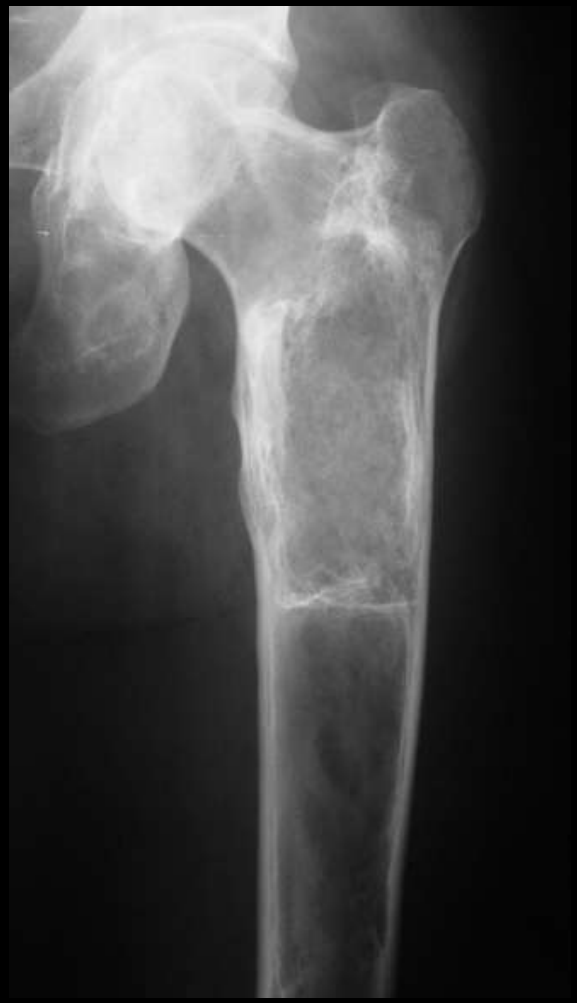


Fibrous Dysplasia
General

- Unknown pathogenesis
- Medullary bone replaced by fibrous tissue
- Diagnosed 3-15 years
- Mono or Polyostotic
  - Polyostotic tends to be unilateral
  - If bilateral then asymmetric
- Femur, Pelvis, Skull, Mandible, Ribs, Humerus
  - Commonest expansile rib lesion

Fibrous Dysplasia
Radiology

- Cyst like lesion
- Meta or Diaphysis, Epiphysis after fusion
- +/- Expansion
- +/- Endosteal scalloping
- No periosteal new bone
- Thick sclerotic border “rind sign”
- Ground glass matrix +/- irregular calcifications



Fibrous Dysplasia
Radiology

- Shepherds crook deformity of proximal femur
- Growth disparity
- Accelerated bone maturation

Fibrous Dysplasia
Radiology - Skull

- Mixed lucencies and sclerosis
  - Convexity of calvarium and floor of anterior fossa
- Leontiasis ossea
  - Sclerosing form affecting face
- Cherubism
  - Lytic expansion of mandible and maxilla

Fibrous Dysplasia Associations

- Sexual precocity + Café au lait
- 30% of polyostotic form
- McCune Albright syndrome

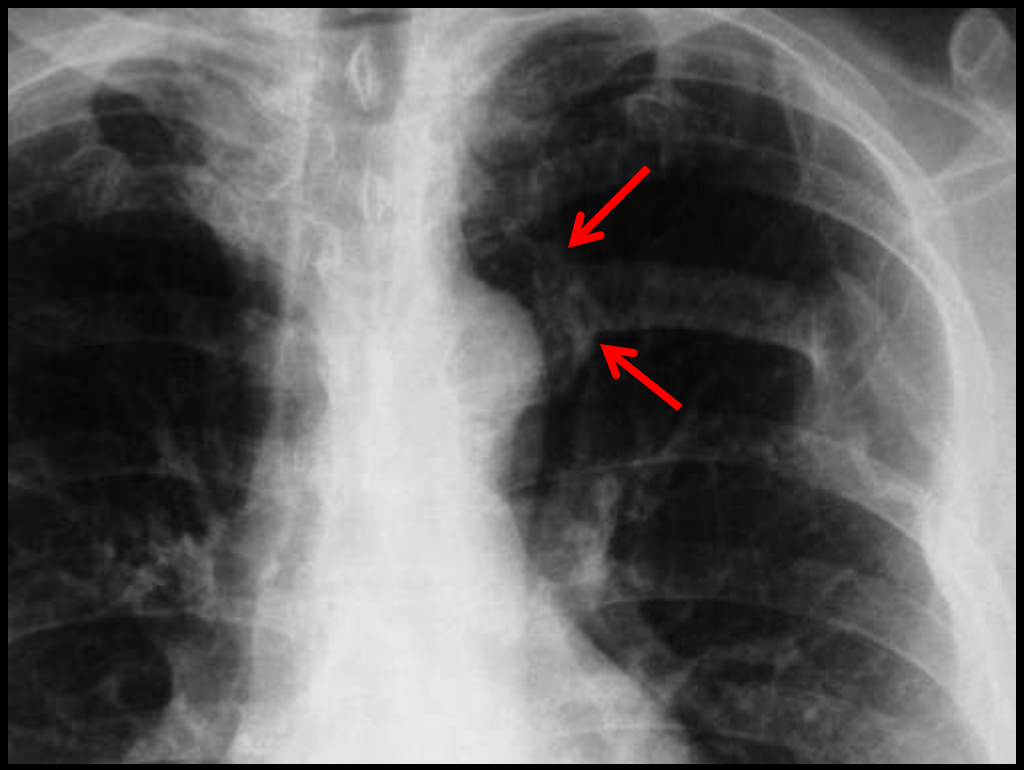
- Acromegaly

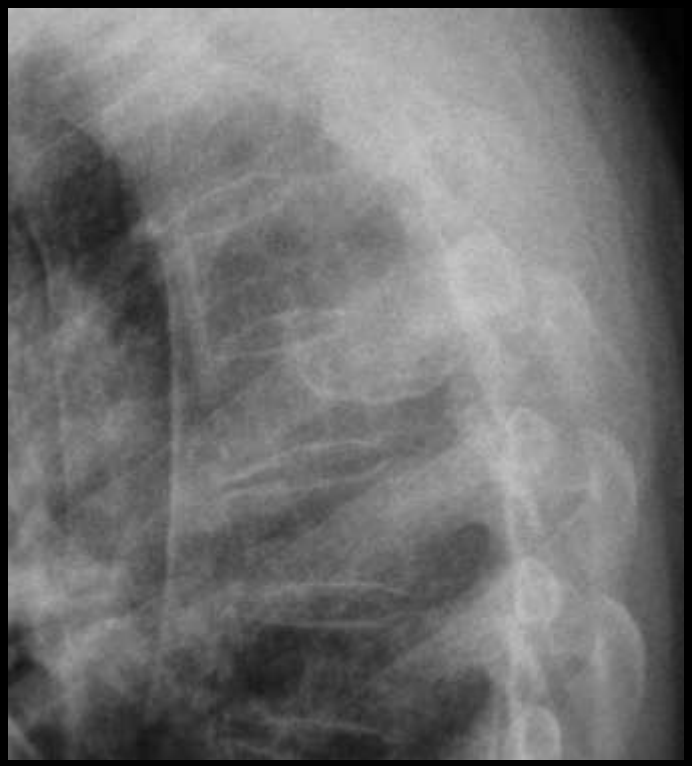
- Cushing’s syndrome

- Gynaecomastia

- Parathyroid hyperplasia

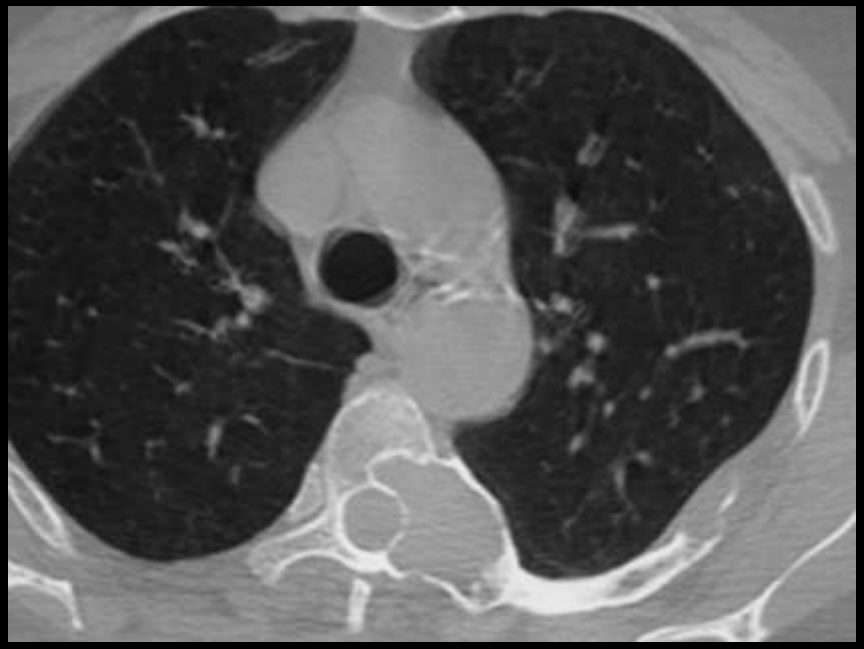
Fibrous Dysplasia

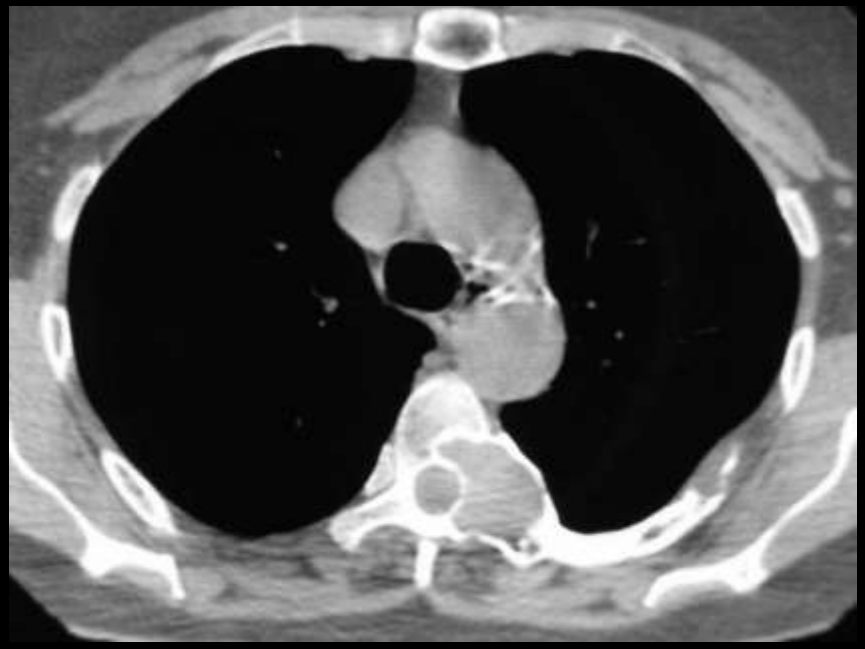




67F Lump

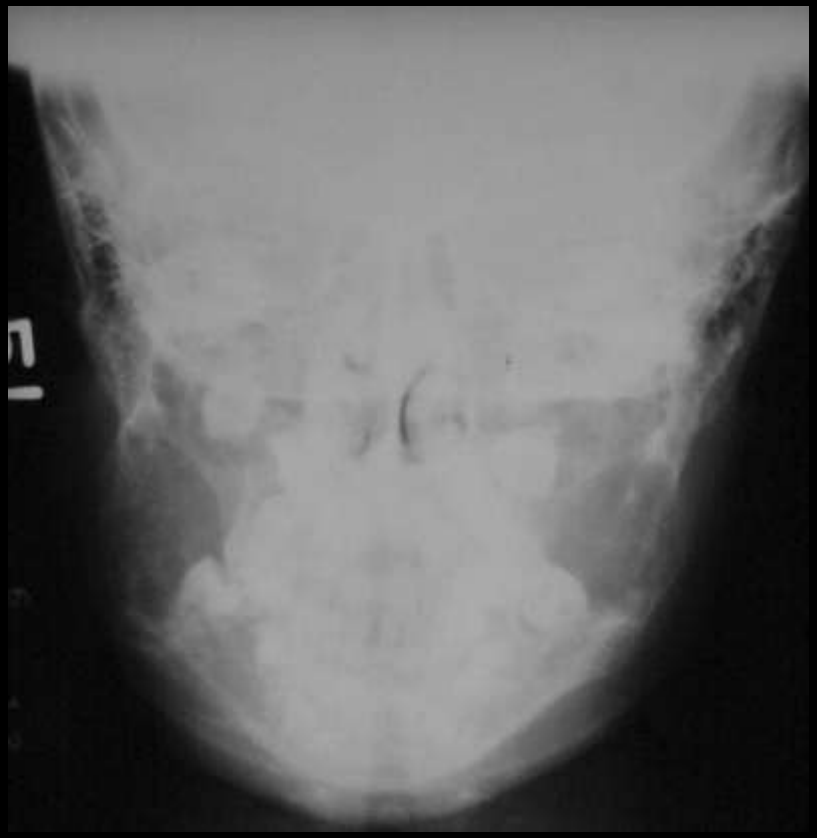
Fibrous Dysplasia

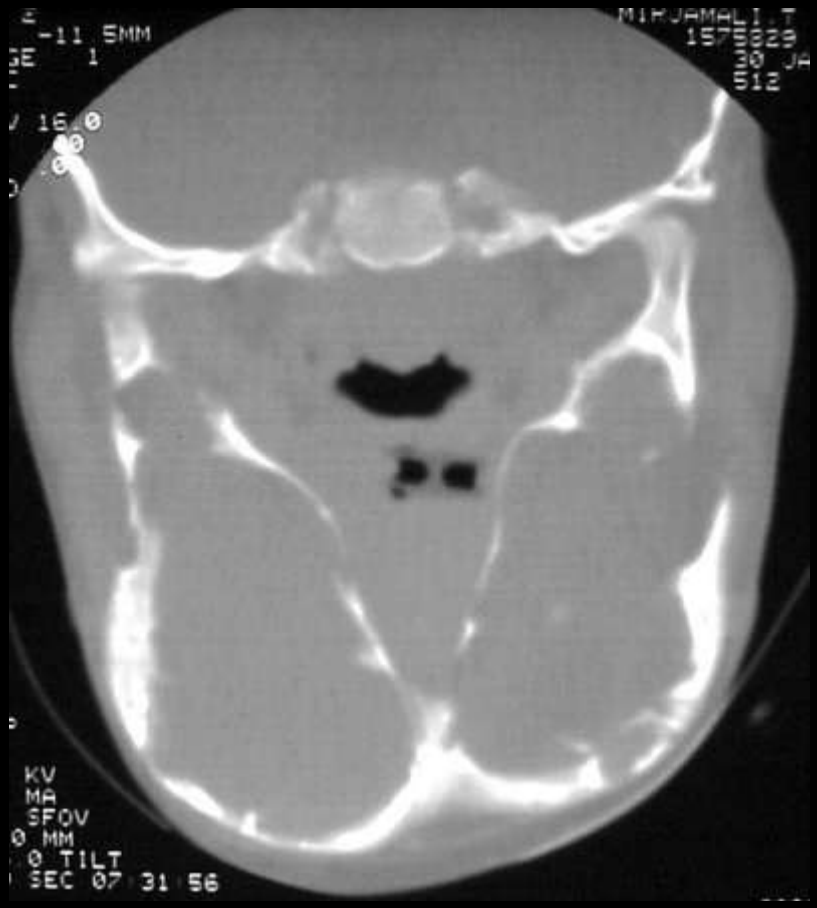




67F Lump

Fibrous Dysplasia - Cherub

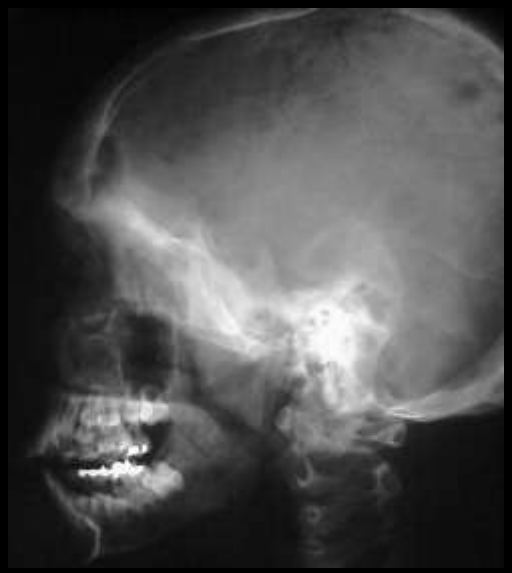


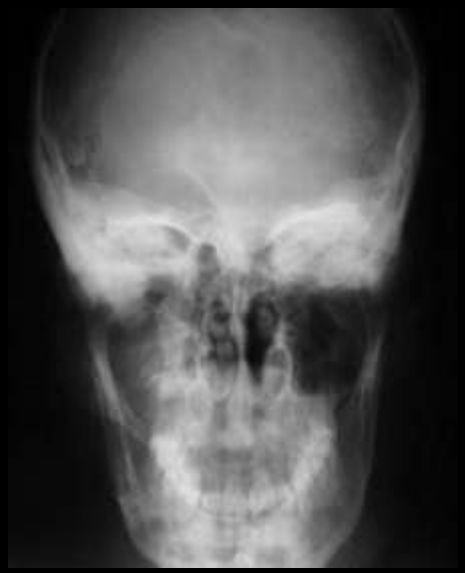


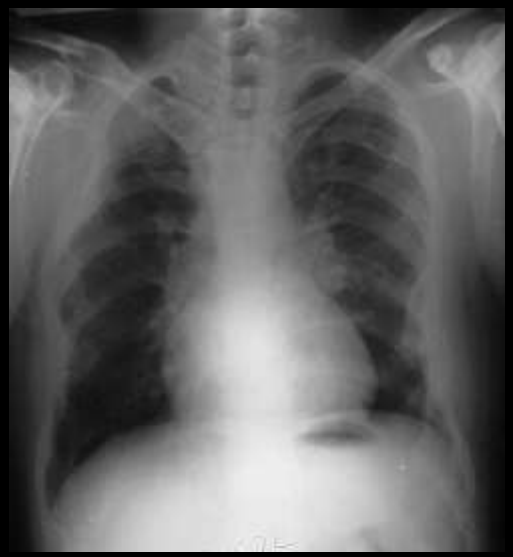
Fibrous Dysplasia

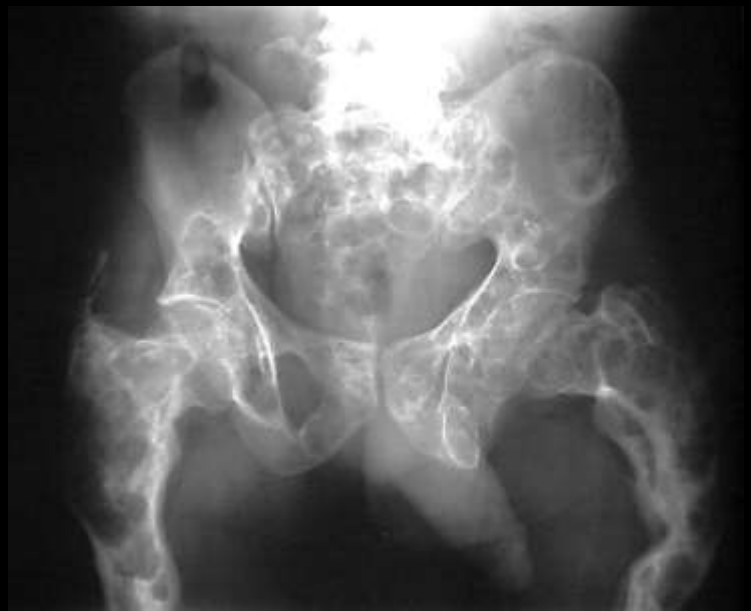
- Common
- Hamartomatous fibro-osseous metaplasia
- 70% monostotic
- Polyostotic tends to be unilateral
- Usually expansile
- Shepherds crook, ground glass
- Any bone, but spine unusual

Polyostotic fibrous dysplasia





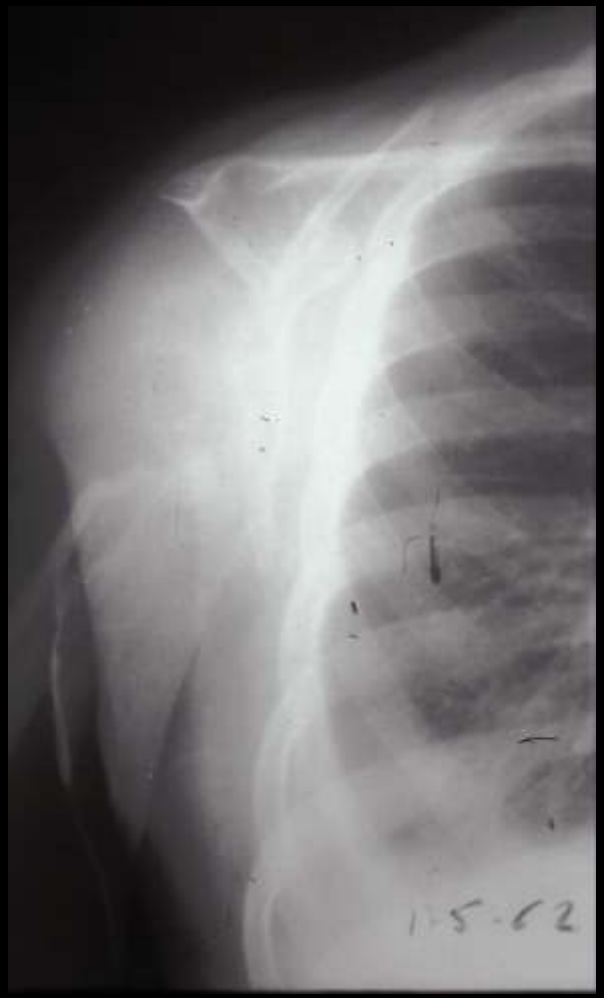


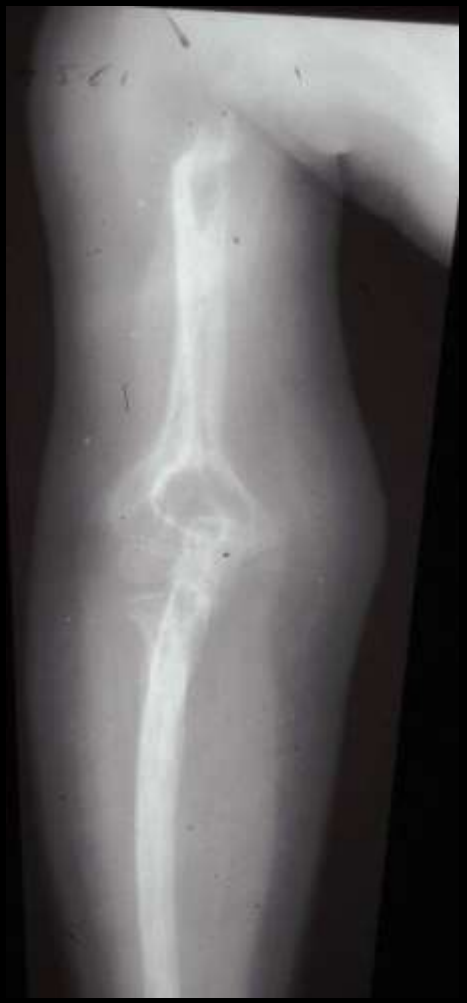


Acroosteolysis

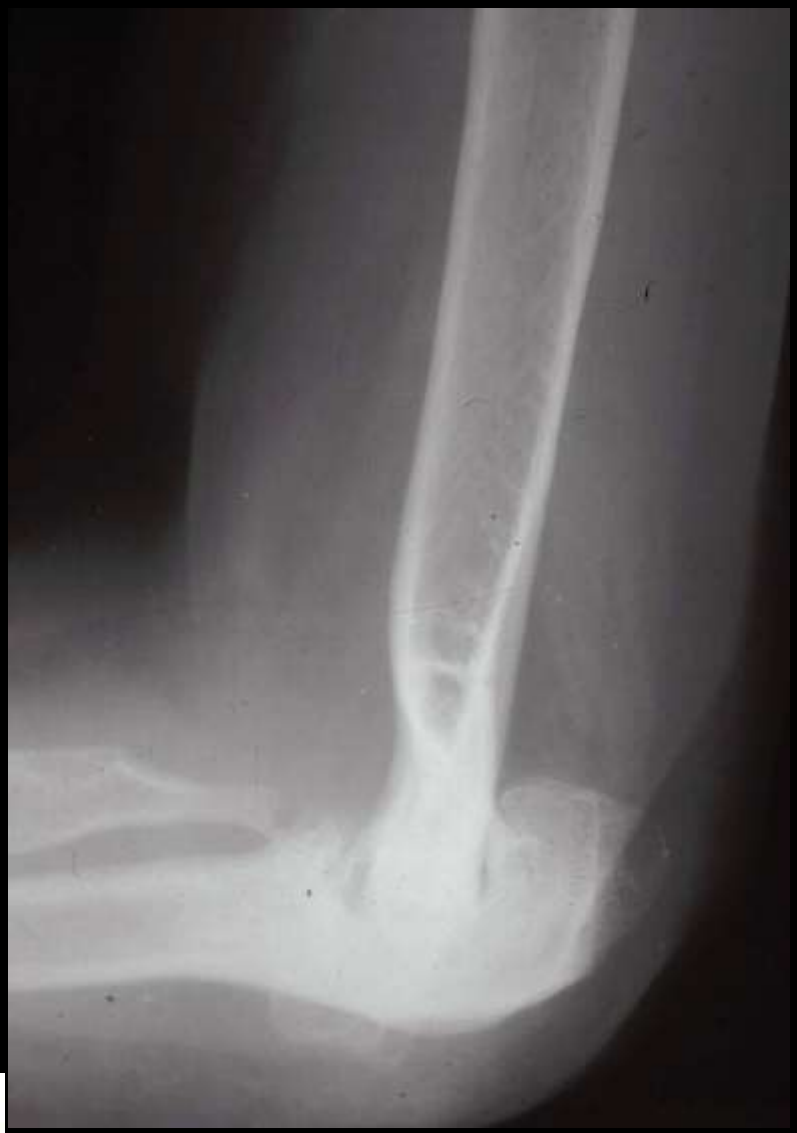
- Tuft
  - CVD, Psoriatic, Neuropathic, Thermal, Trauma, HPT, Porphyria, Epidermolysis bullosa, Phenytoin toxicity, Subungal exostosis, Snake venom
- Middle third
  - HPT, Hajdu Cheney, PVC
- Periarticular
  - Psoriatic, Erosive OA, HPT, Thermal injury, Scleroderma, Multicentric reticulohistiocytosis

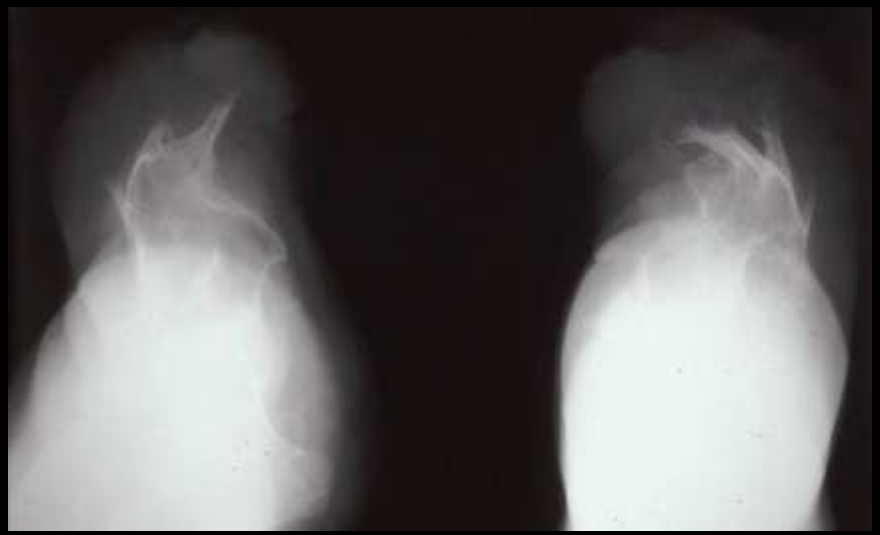
Massive osteolysis

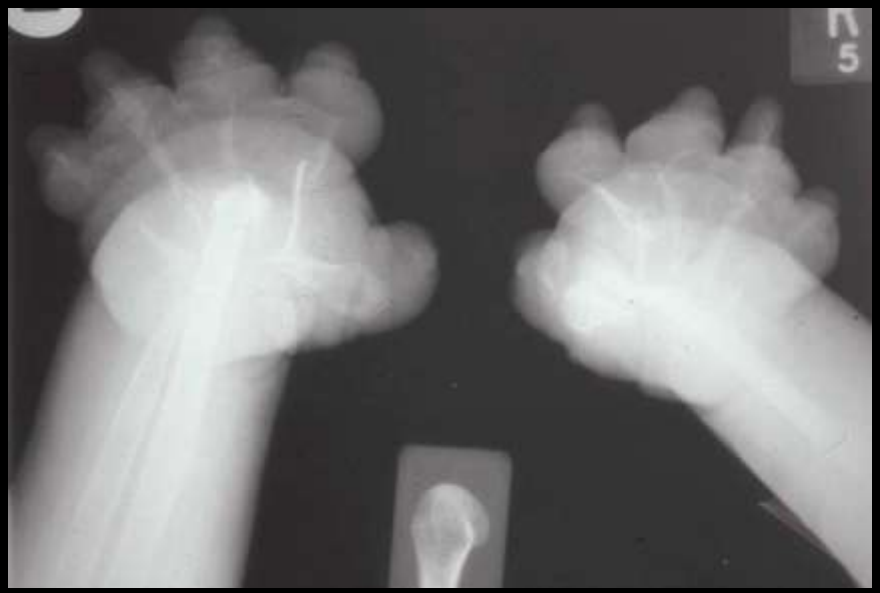




Familial Osteolysis



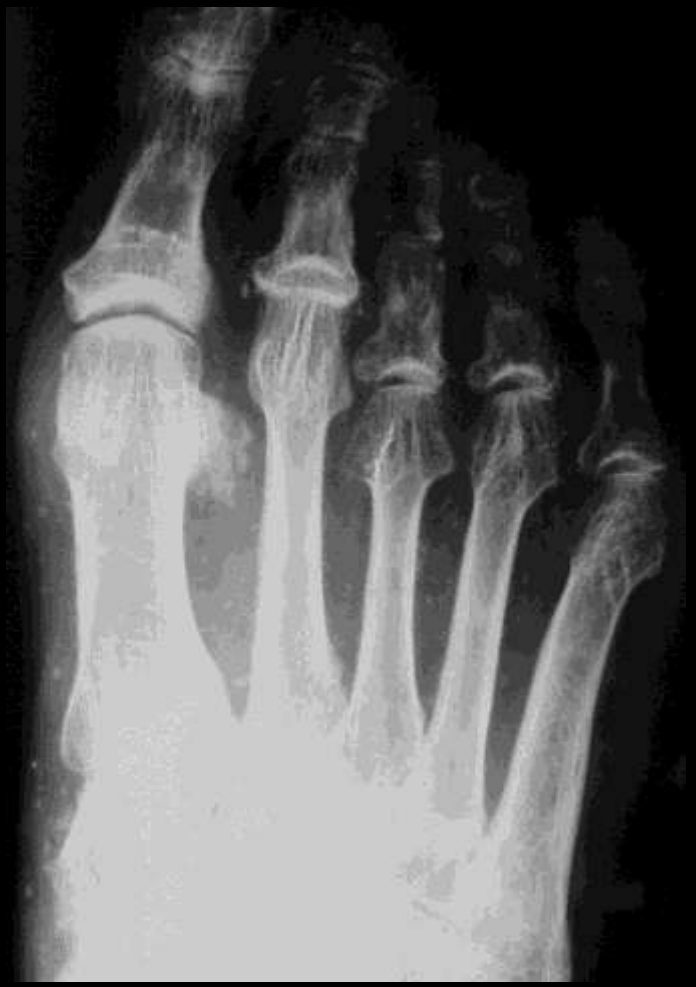




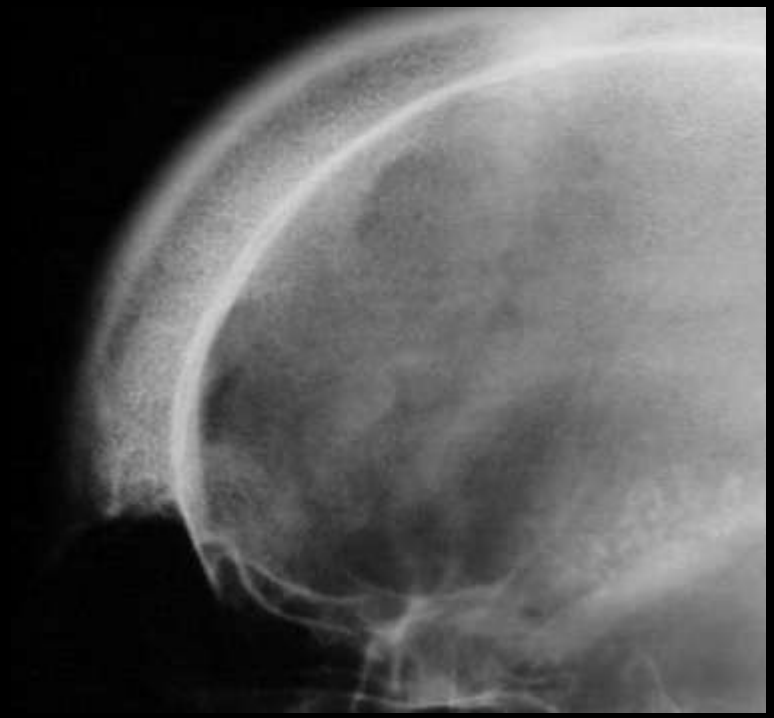
Pseudohypoparathyroidism

- X-linked, renal and skeletal resistance to PTH
- Decreased Ca, normal/increased PTH
- Short, retarded
- Brachydactyly 1,4,5 MC
- Ca basal ganglia, skin, SubQ

Pseudohypoparathyroidism



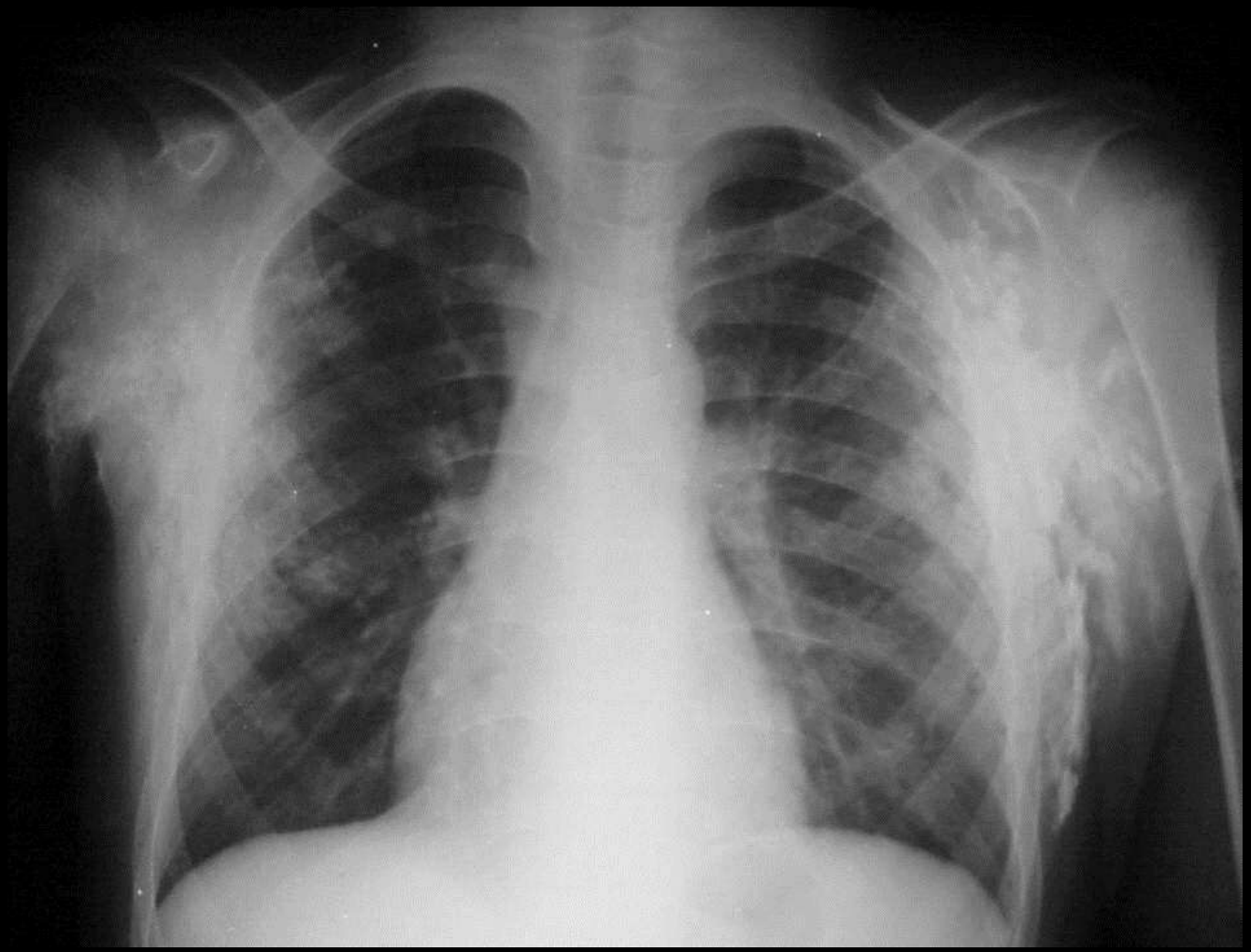
Pseudohypoparathyroidism



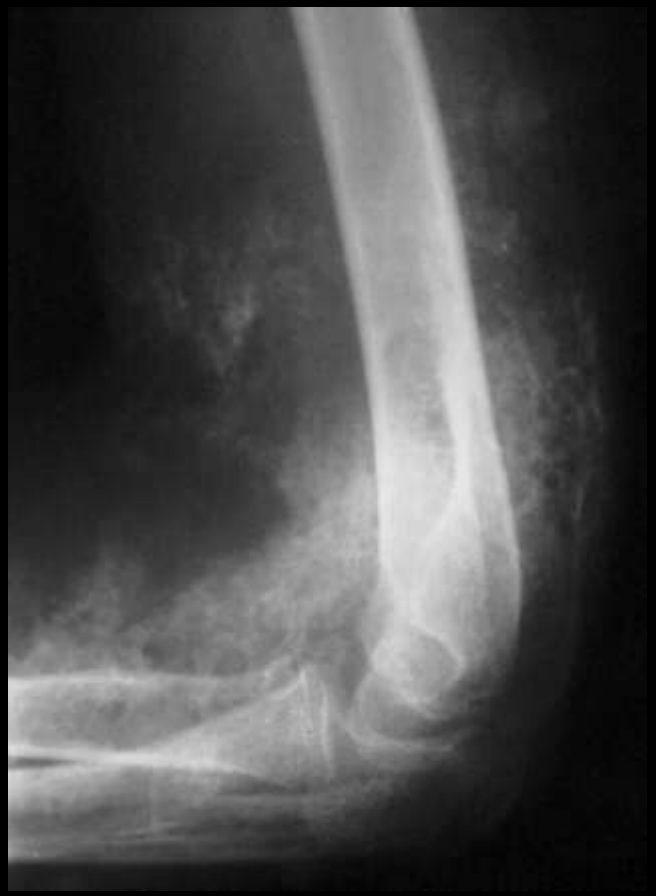
Dermatomyositis

- Damaged chondroitin sulfate
- Atrophy, oedema, necrosis of muscle
- 30-60, F>M
- Calcification extremities and girdles
- Pointing of tufts
- Ass. Malignancy, lung, kidney, ovary, breast

Dermatomyositis



Dermatomyositis



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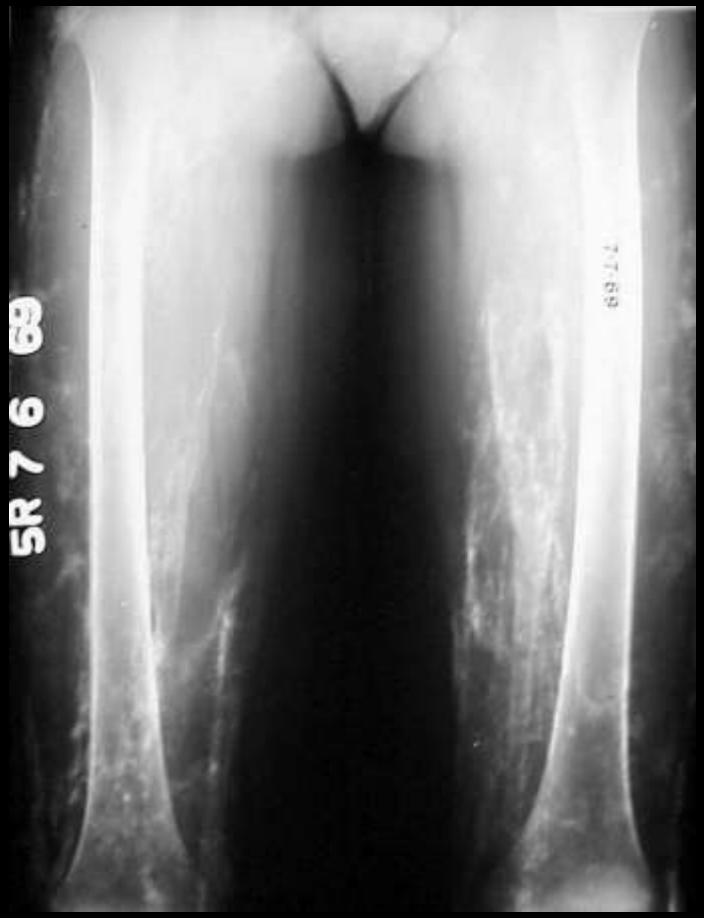

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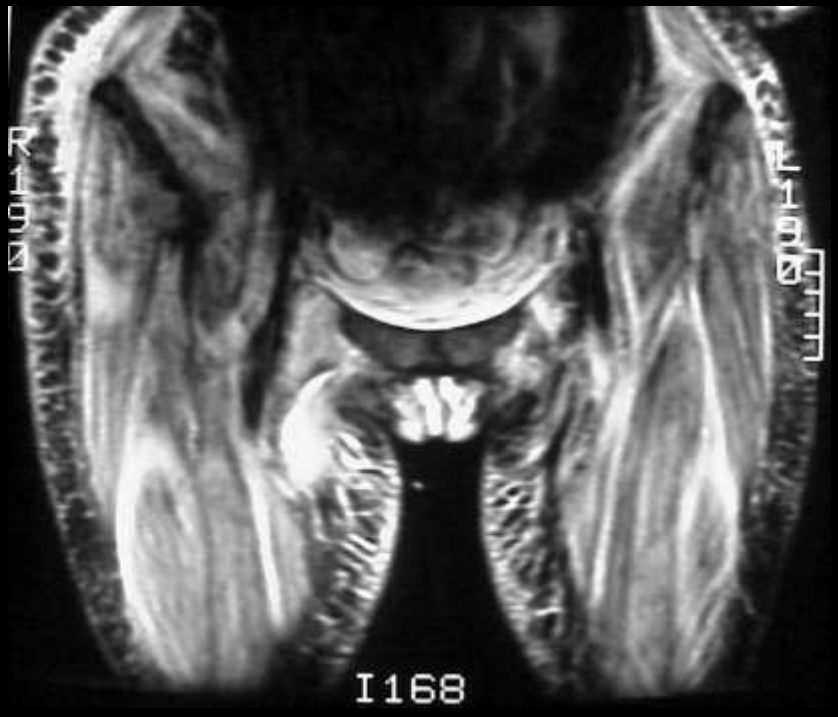
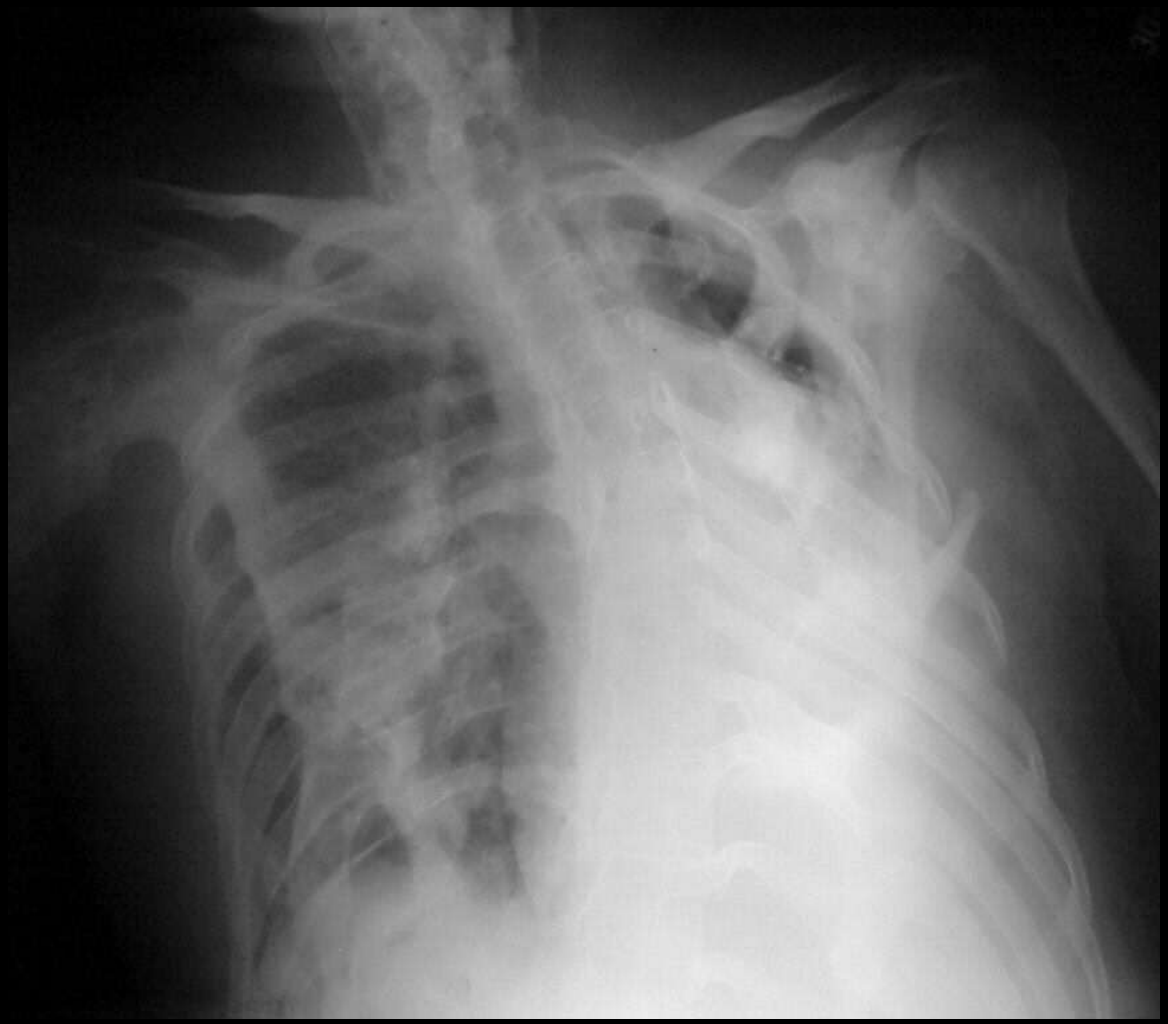
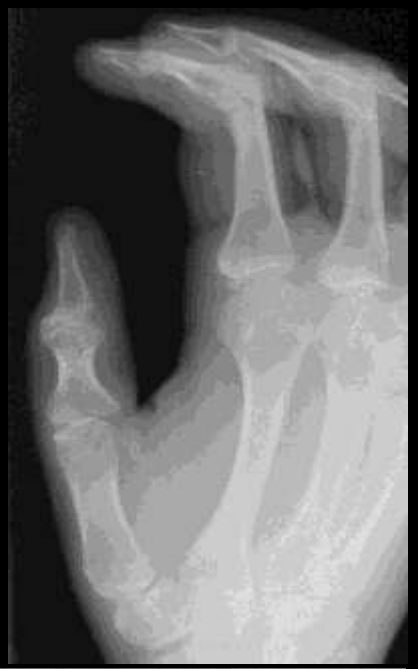


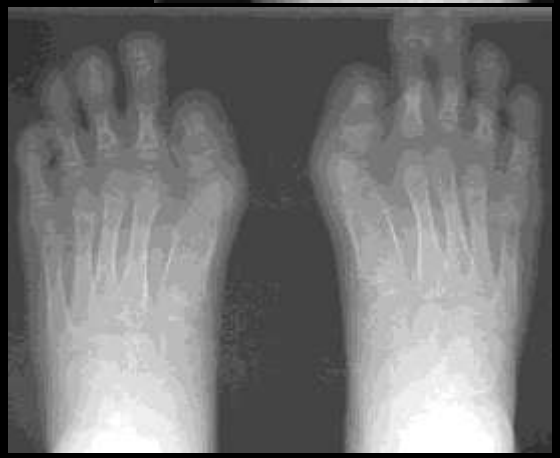
Fibrodysplasia ossificans progressiva

- MOP / Stone man
- Rare, AD, sporadic
- Presents in childhood
- Stiffness, Heterotopic ossification
- Malformed fingers and toes
- Bone morphogenic protein (BMP) signaling pathway problem

Fibrodysplasia ossificans progressiva



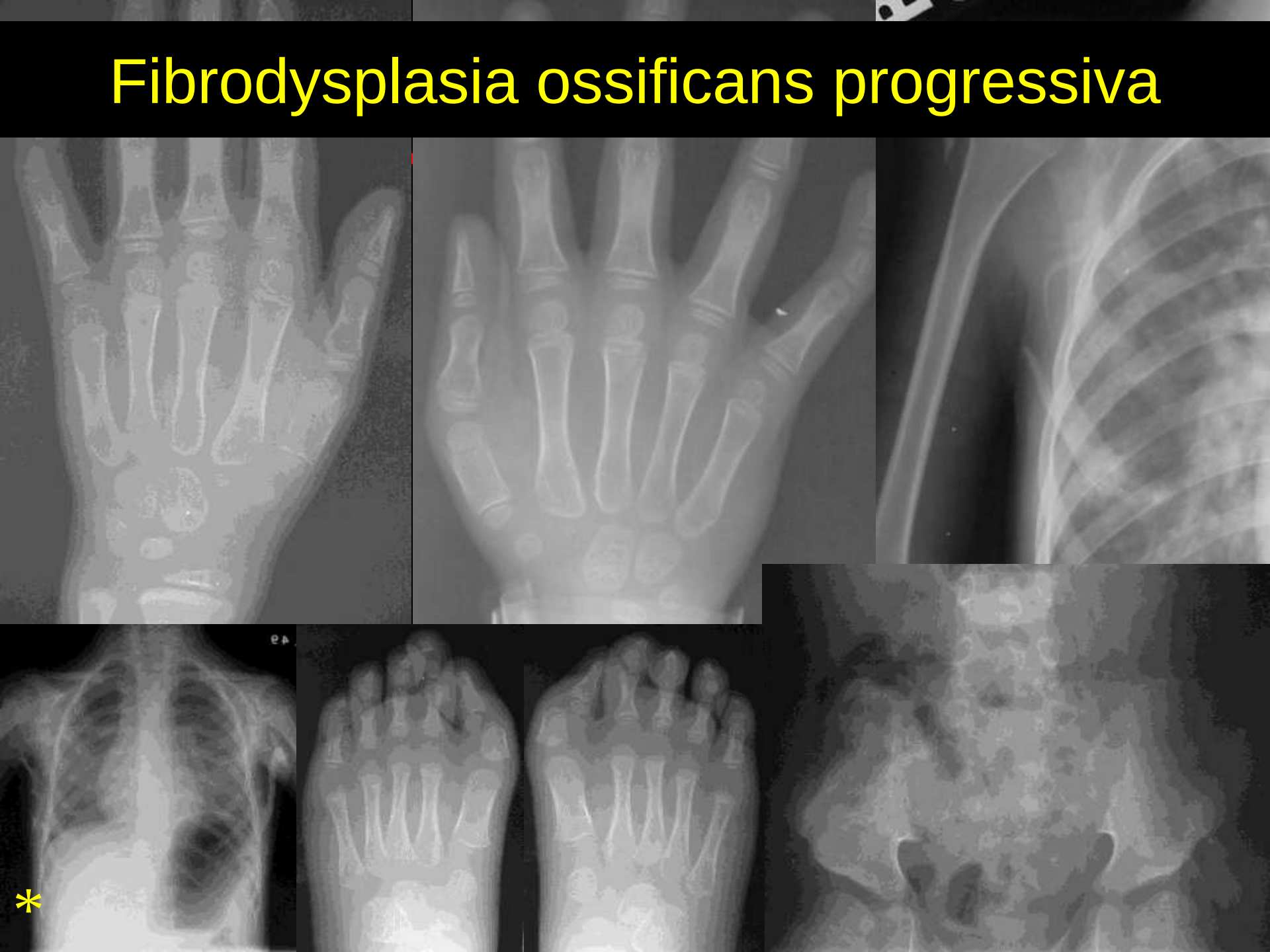




*

37F acute torticollis, stiffness and shortness of breath

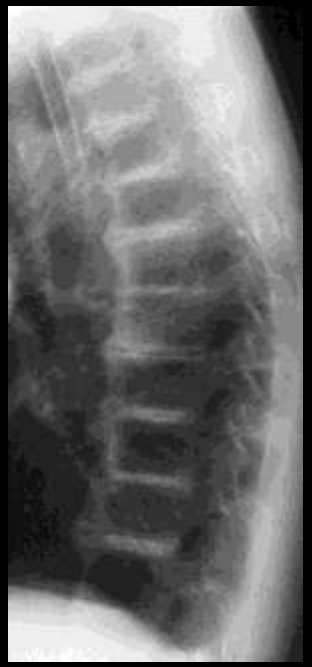
Fibrodysplasia ossificans progressiva

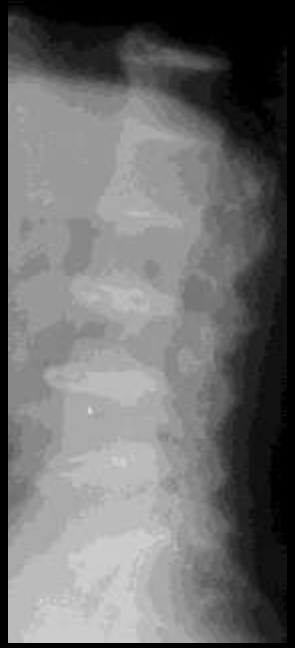


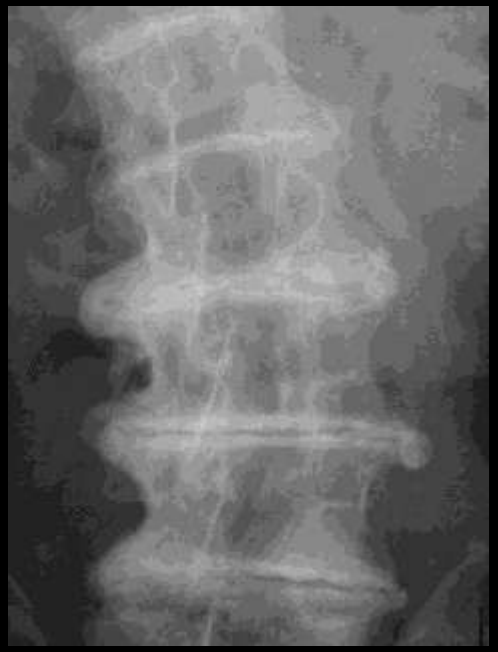
Alkaptonuria / Ochronosis

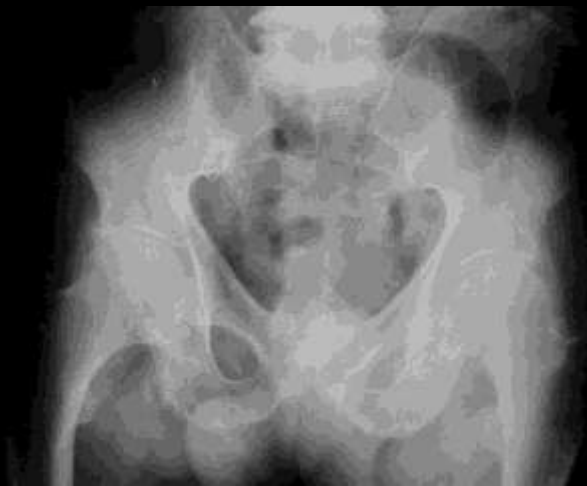
- Absence of homogentisic acid oxidase
- Pigmentation
- Arthropathy
- Osteoporotic with dense disc calcification
- Larger joints show DJD

Alkaptonuria / Ochronosis







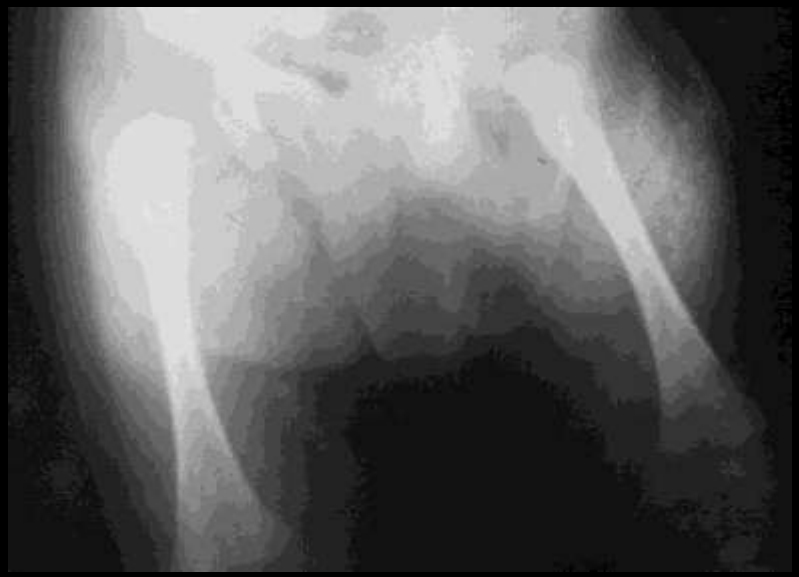


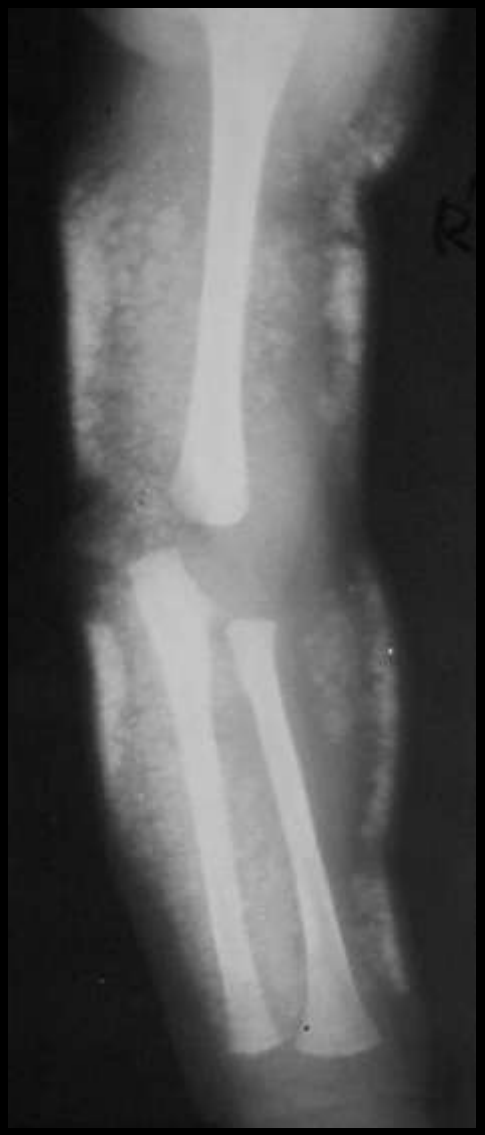
48M joint pains and dark pigmentation on ears

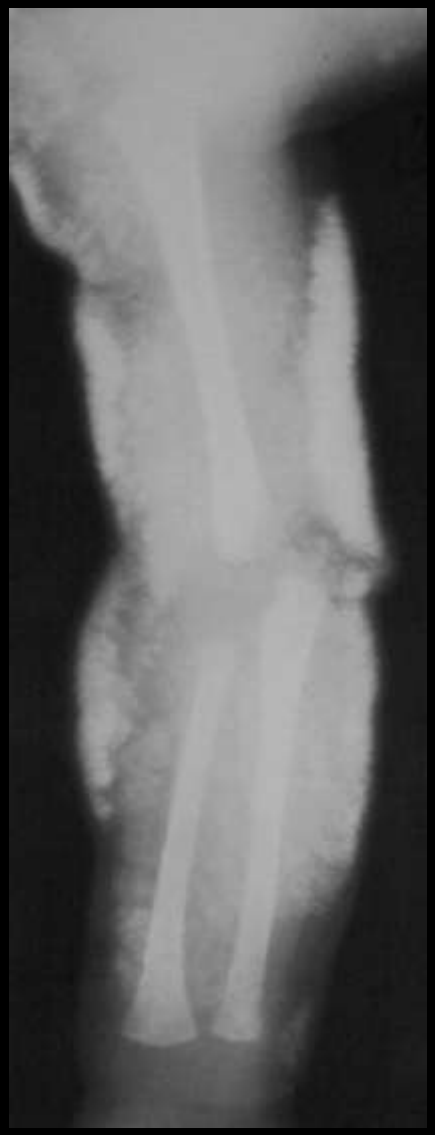
Idiopathic calcinosis universalis

- Rare, unknown cause
- Infants – subcutaneous
- Children - spreads to muscles
- Calcium phosphate and carbonate
- Serum calcium and phosphorous normal
- DDx - DMS, HPT, Calcium gluconate

Idiopathic Calcinosis Universalis

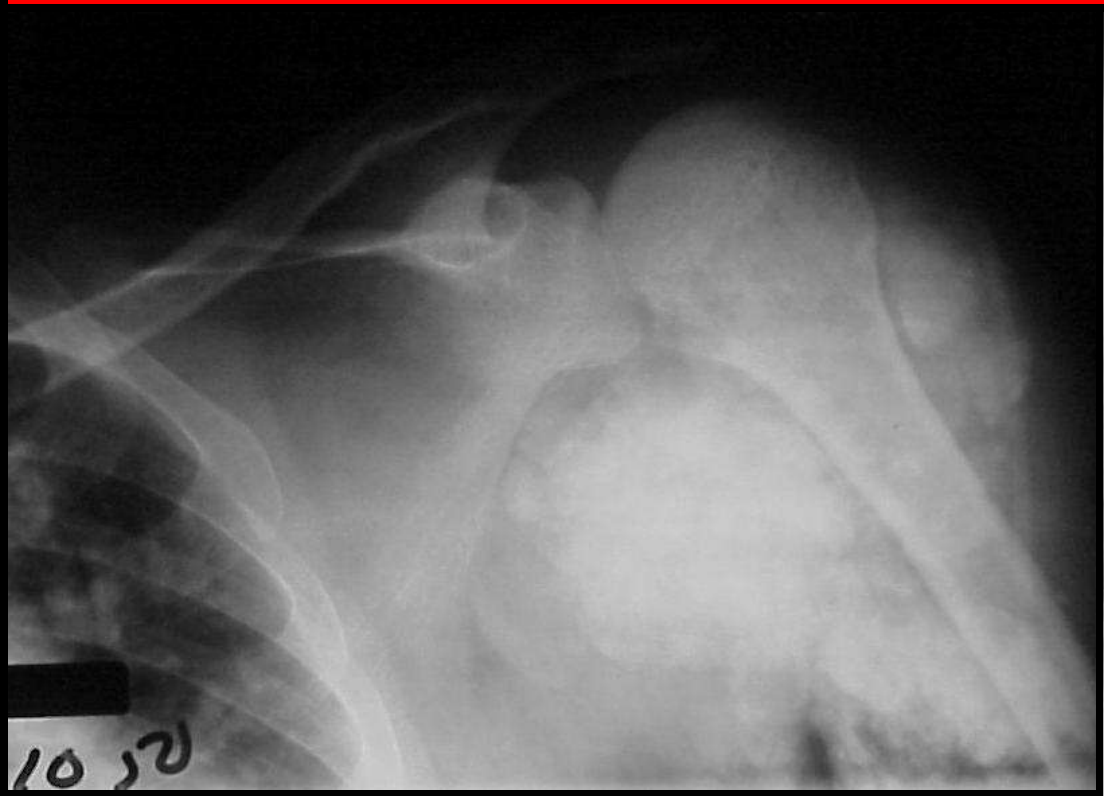


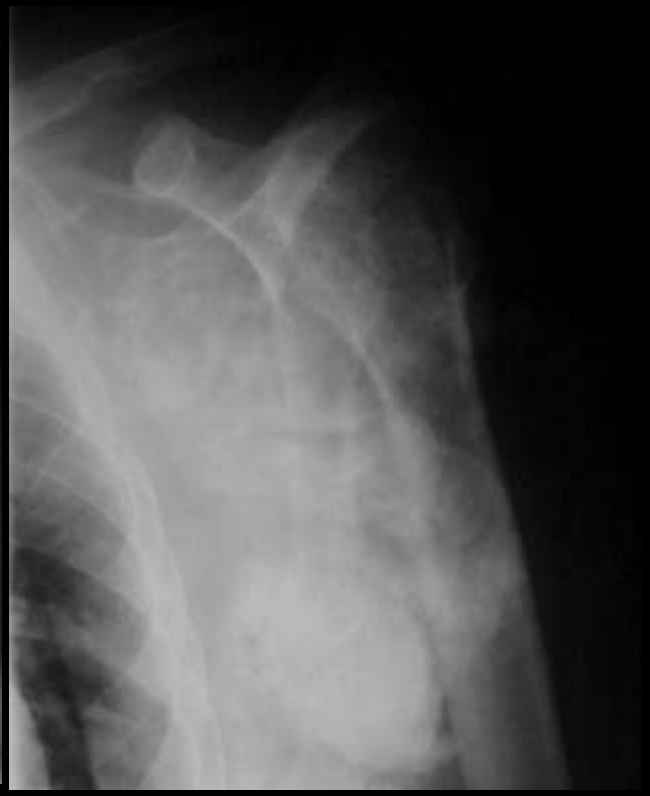




6mM Calcareous nodules

ITC





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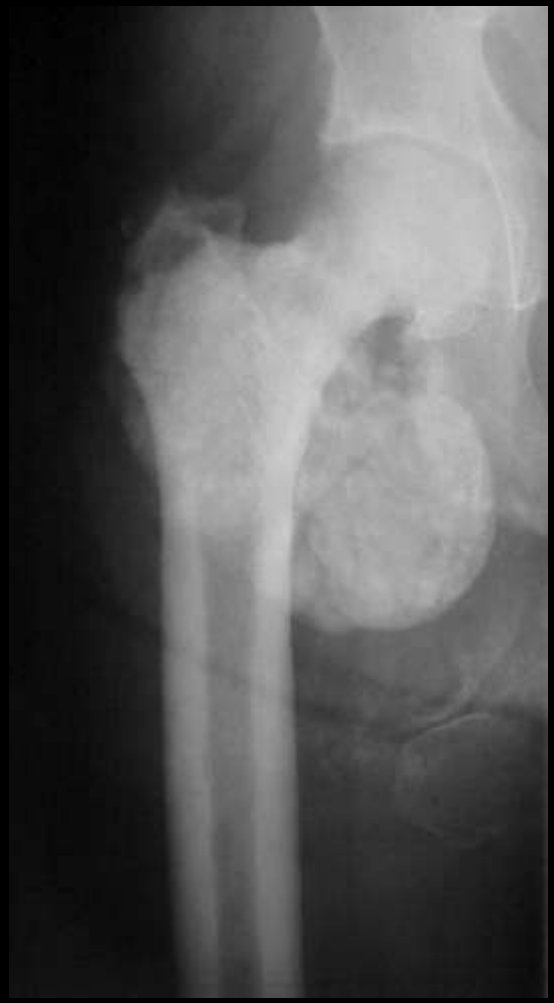
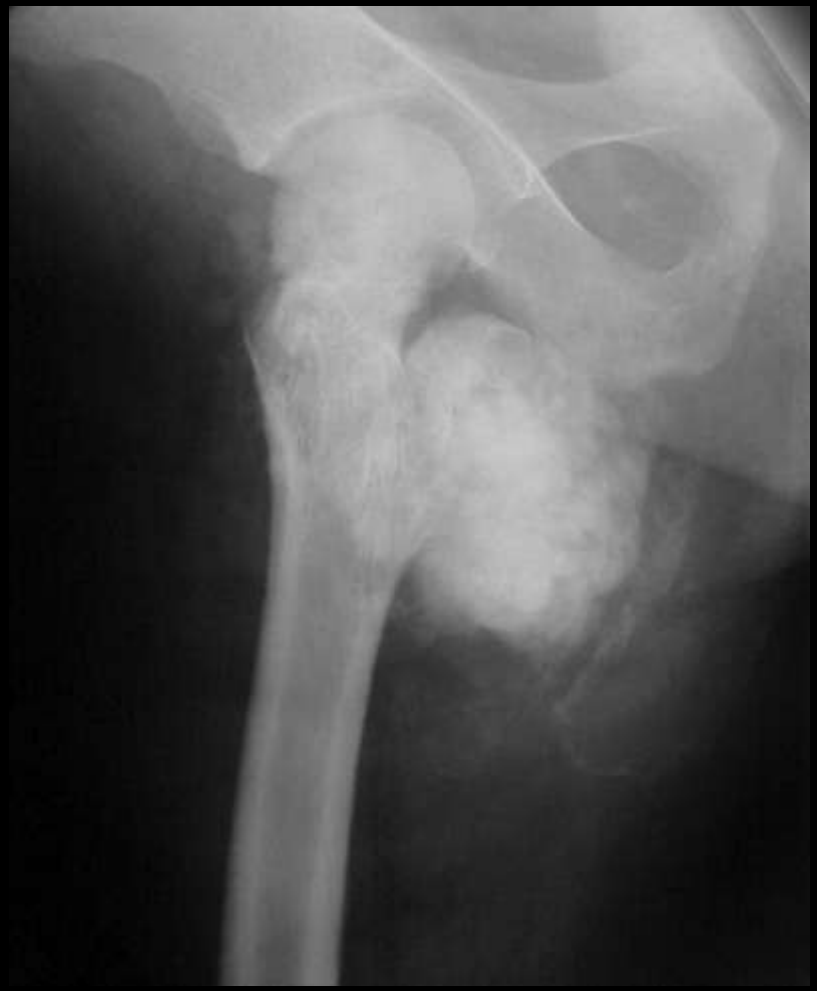
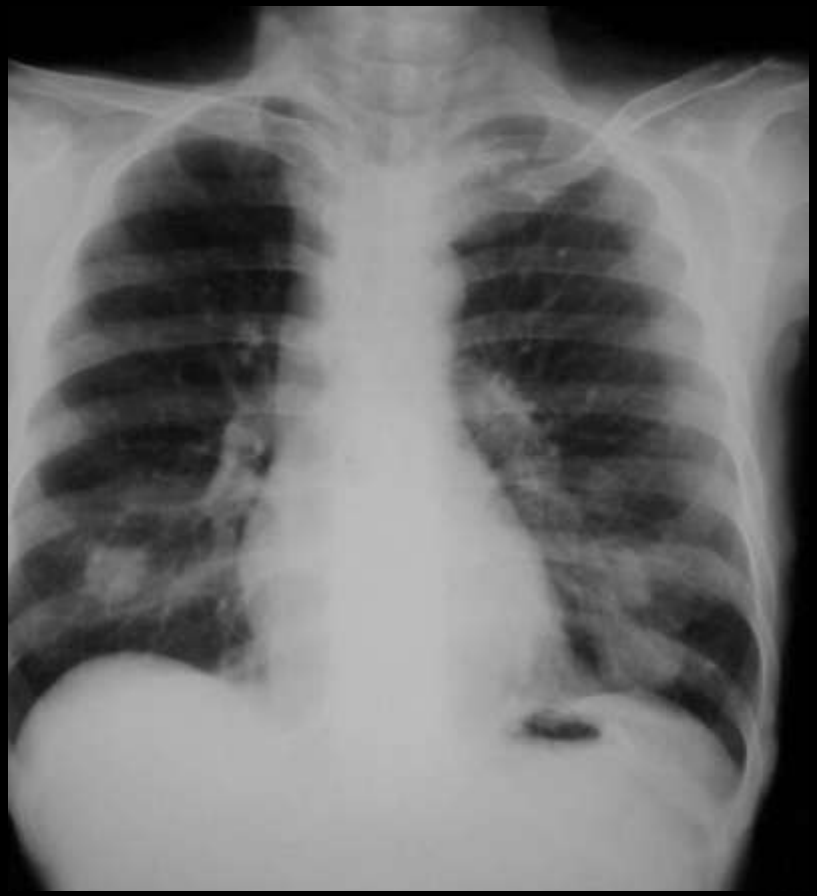
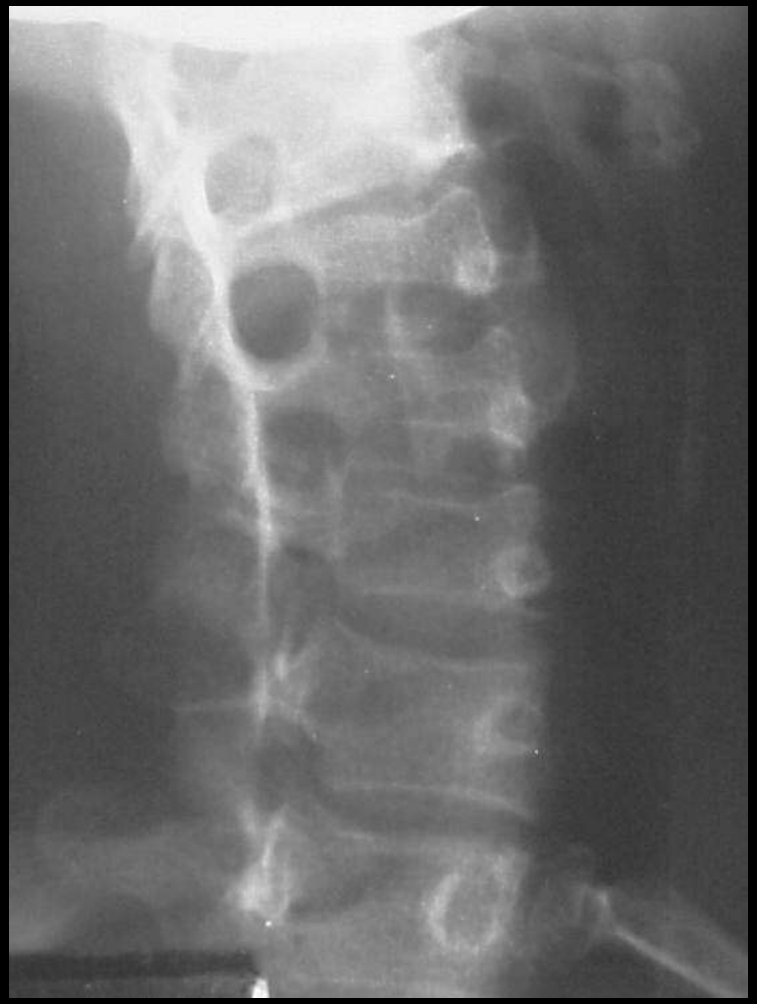


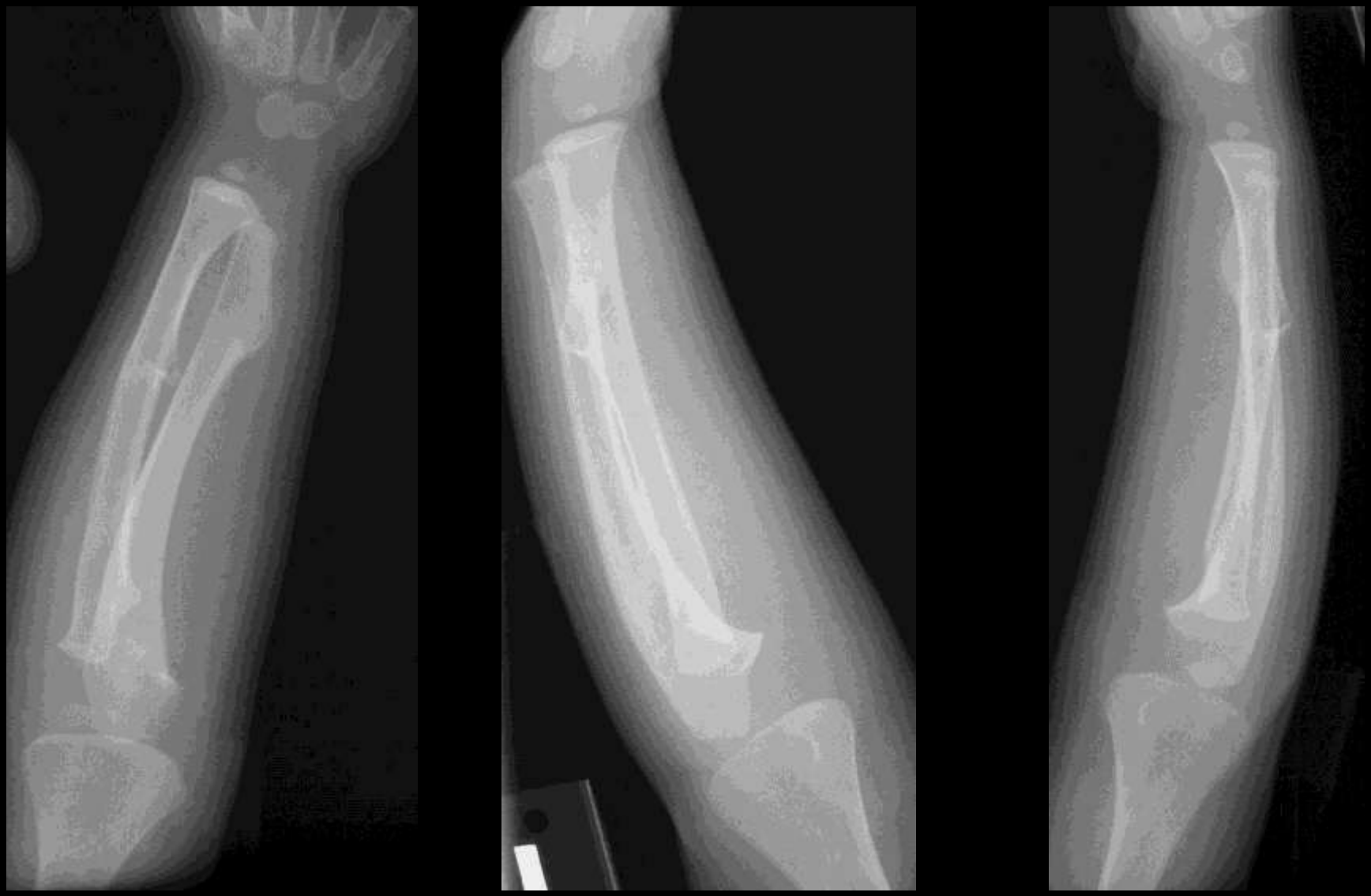


Neurofibromatosis 1



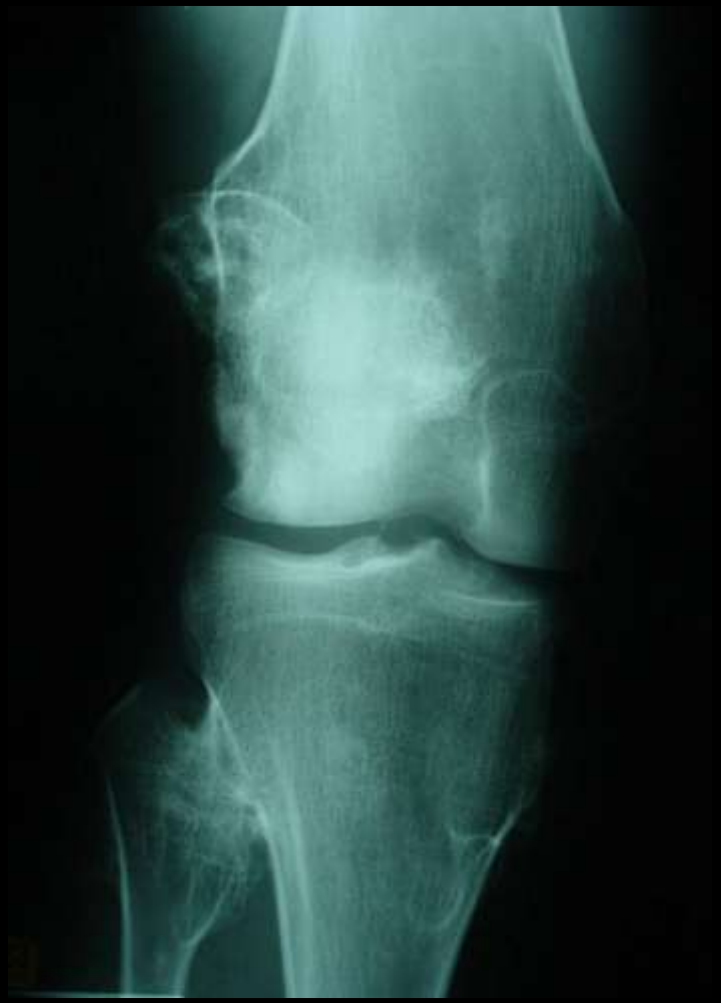


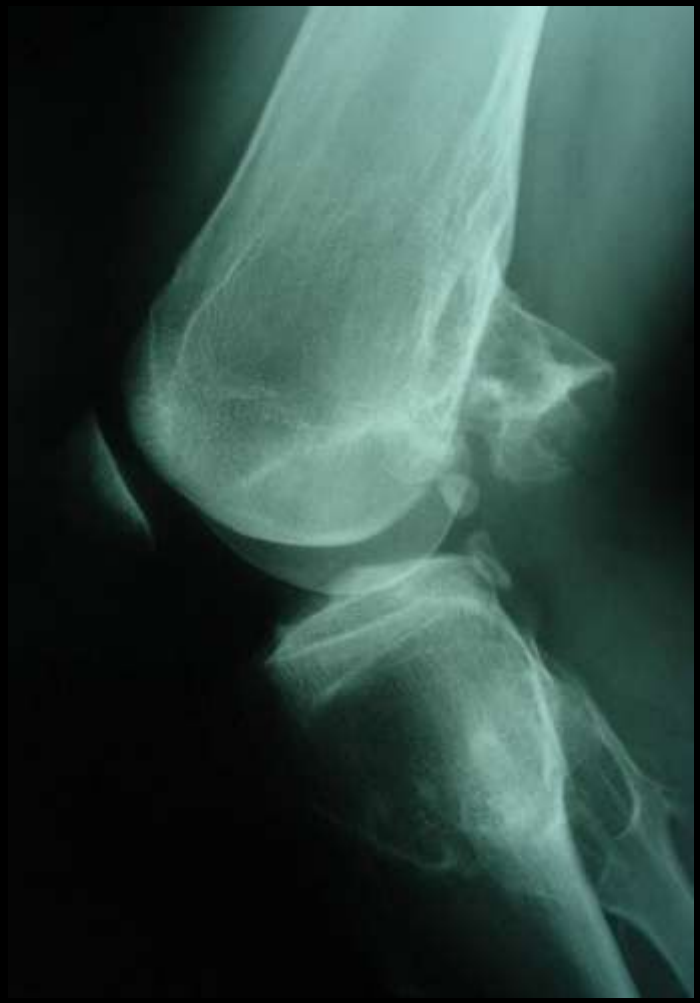
Multiple Hereditary Exostosis



21mF

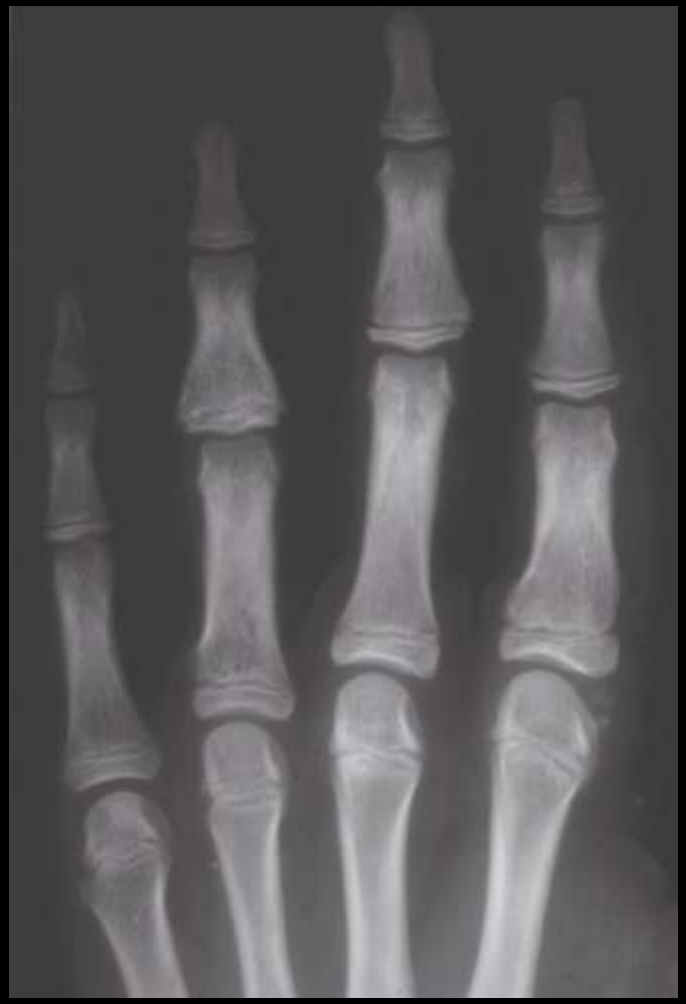
Multiple Hereditary Exostosis

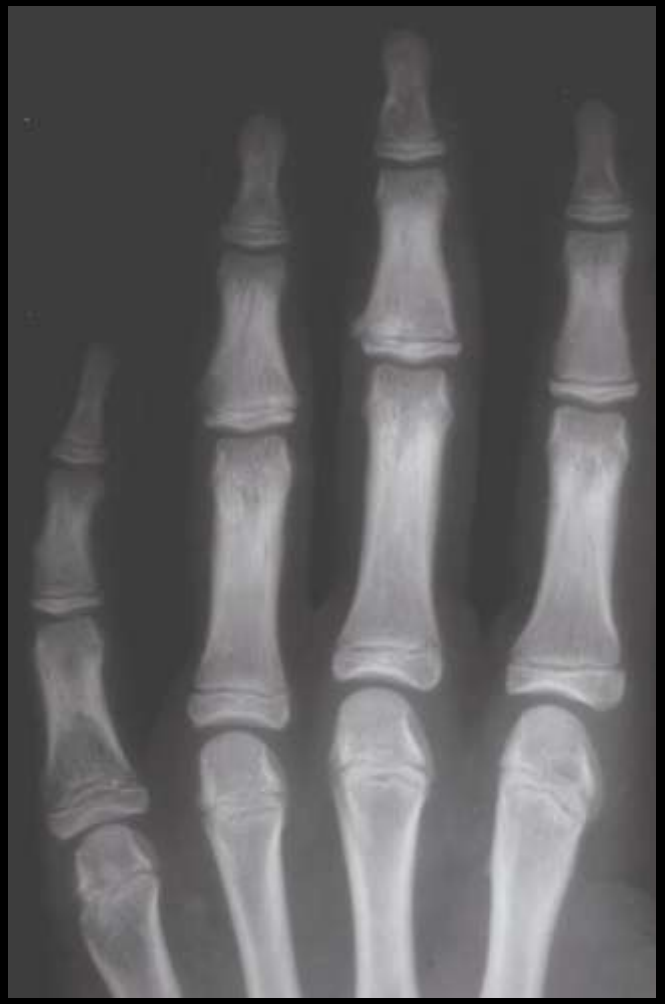




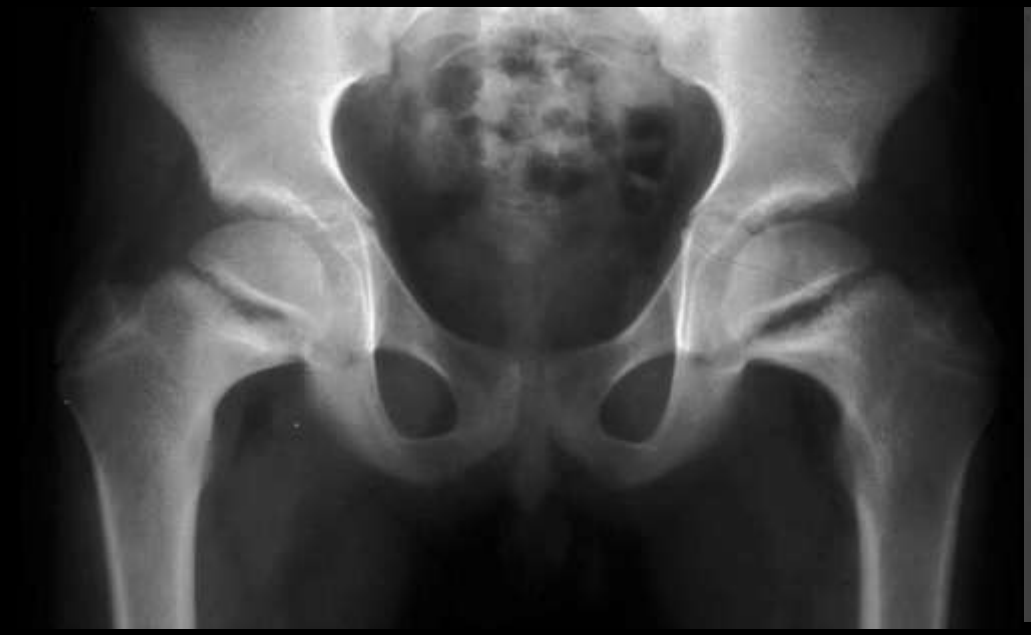
26M

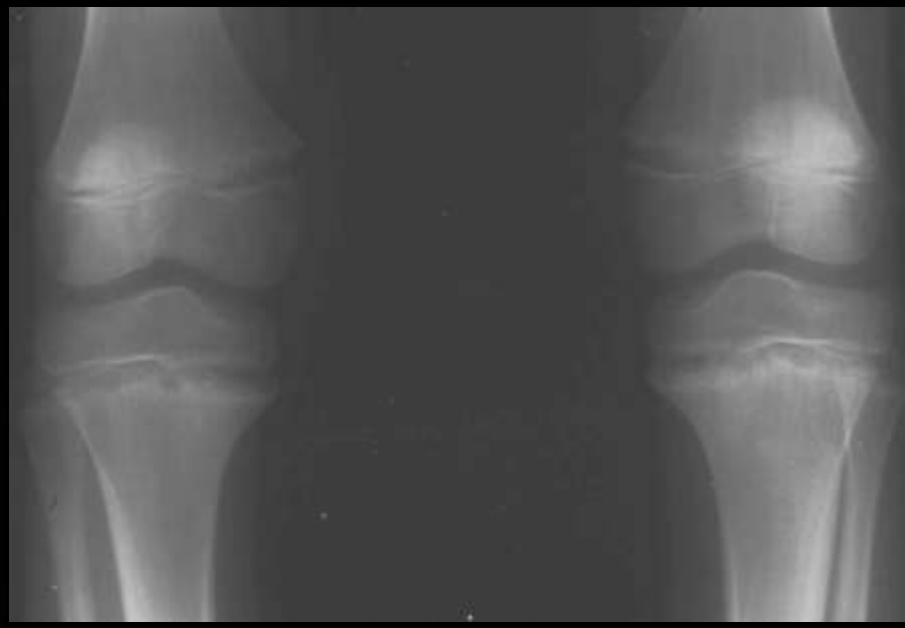
Multiple Hereditary Exostosis





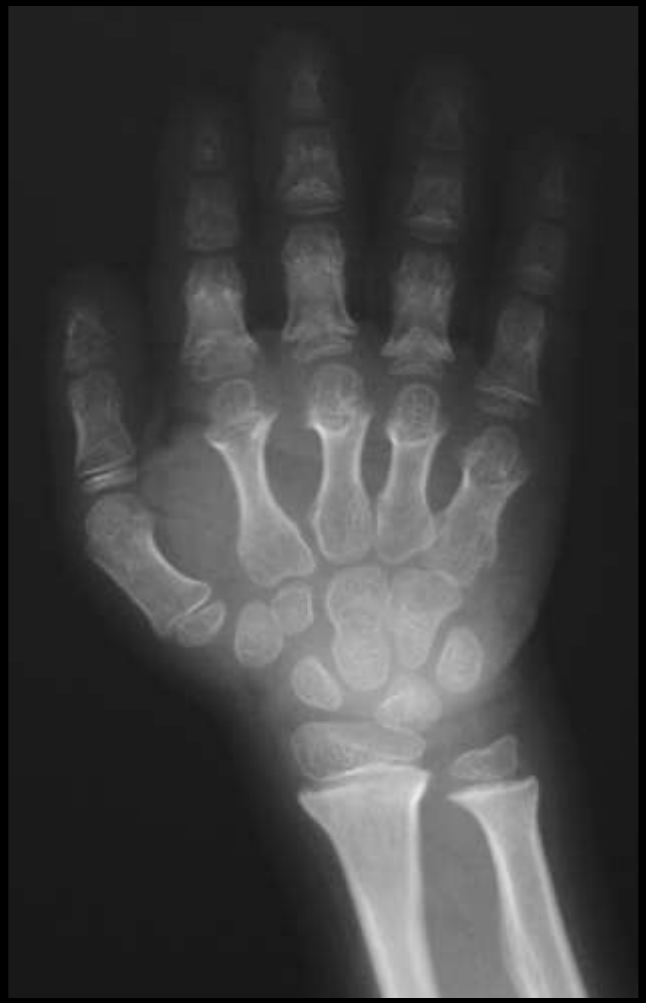
Metaphyseal Dysplasia



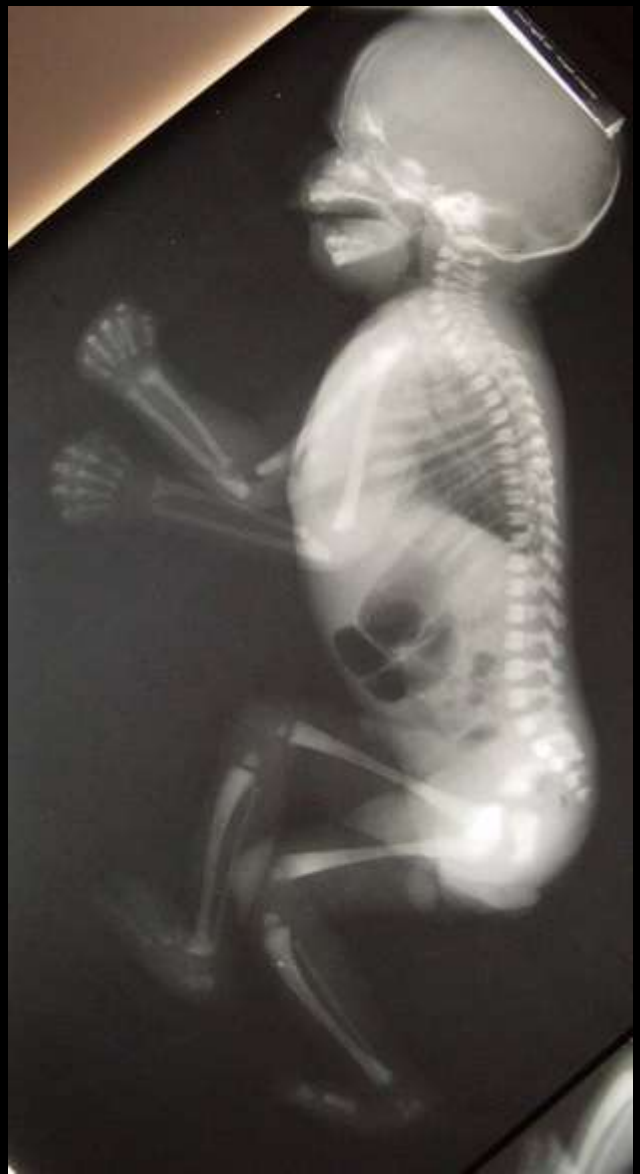


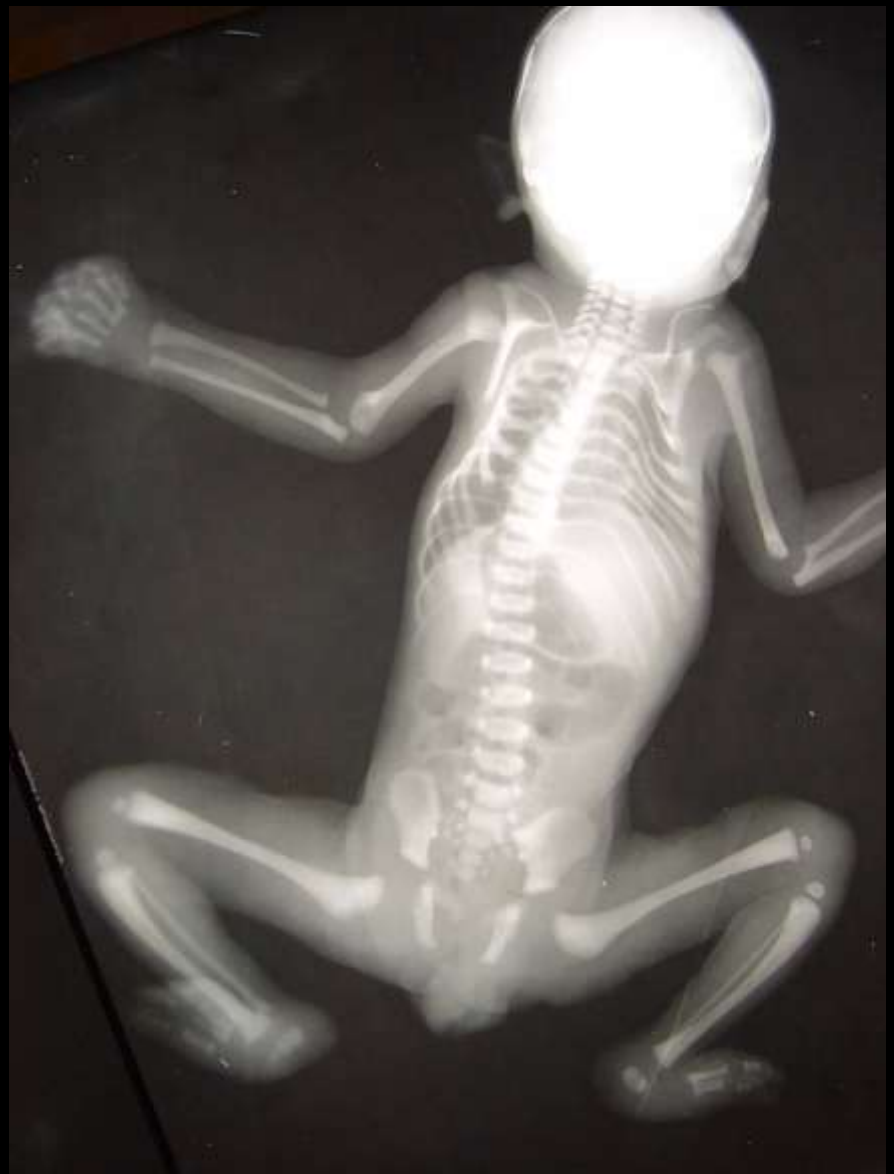
Hips got better

Congenital Metaphyseal Dysplasia



Trisomy 21





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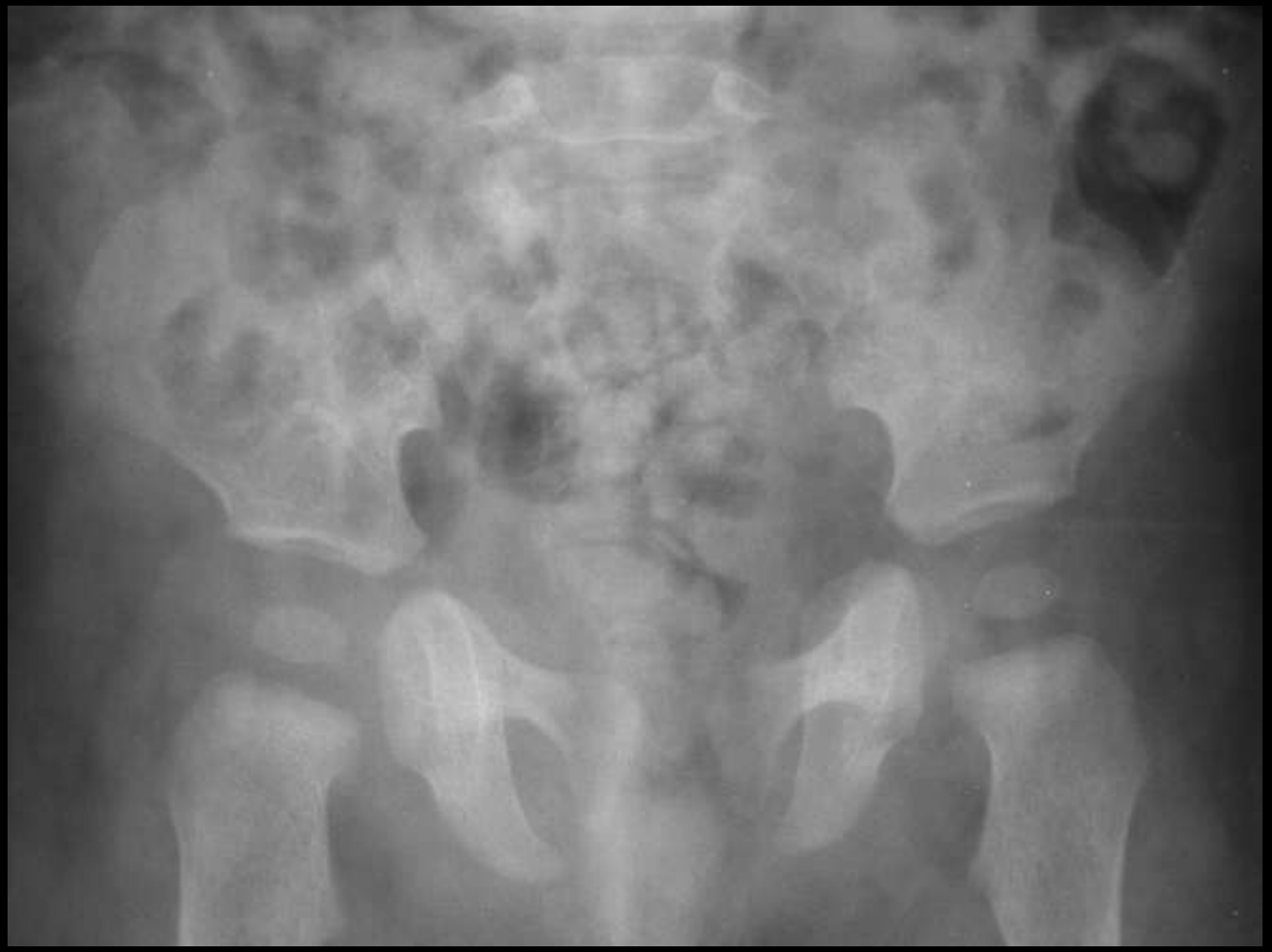

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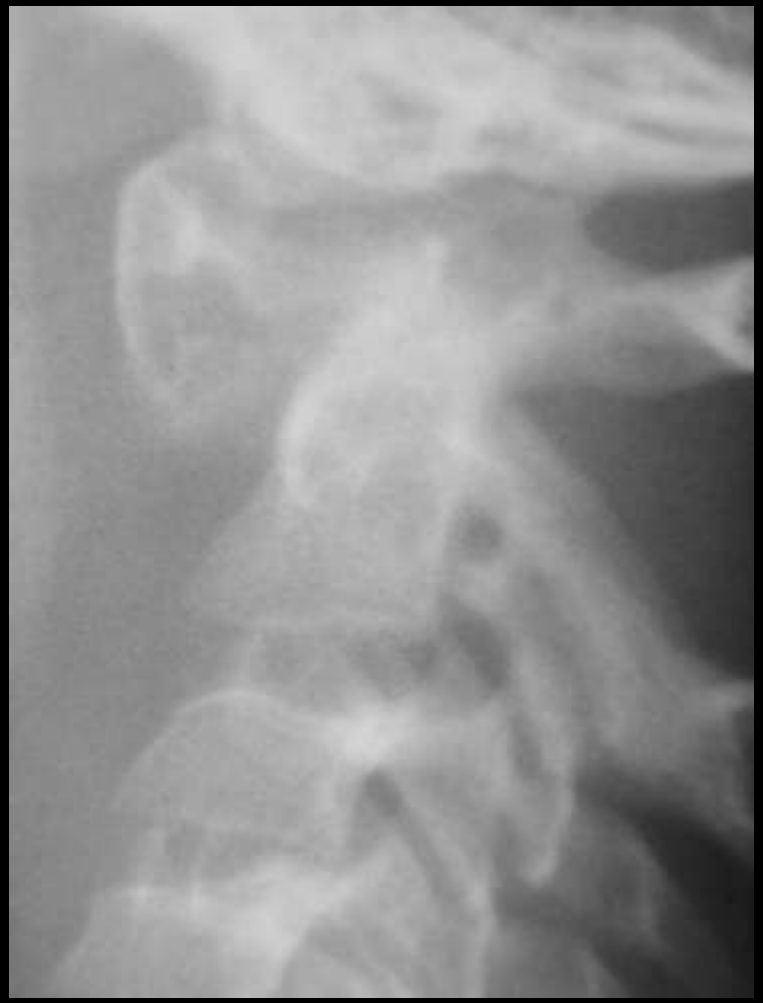
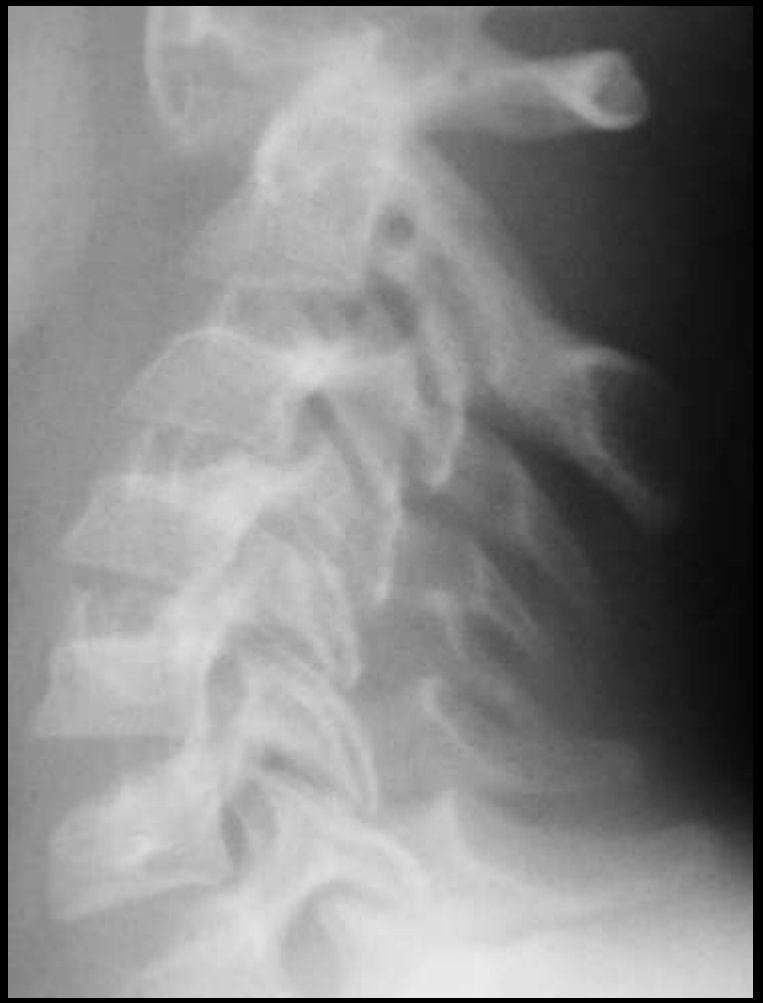




Down's syndrome
General

- Trisomy 21

Down's syndrome
Radiology - Craniofacial

- Brachycephaly and microcephaly
- Hypoplasia of facial bones and sinuses
- Wide sutures with delayed closure
- Wormian bones
- Hypotelorism
- Underdeveloped teeth $\underline{2}$I I$\underline{2}$

Down's syndrome
Radiology – Axial skeleton

- Increased height and decreased AP diameter of lumbar vertebrae
- Atlantoaxial subluxation
- Incomplete fusion of lumbar posterior arches

Down's syndrome
Radiology – Pelvis

- Flared iliac wings
- Small acetabular angles
- Abnormal iliac index
  - Iliac angle + acetabular angle

Down's syndrome
Radiology – Chest

- Eleven pairs of ribs
- Two ossification centers of manubrium

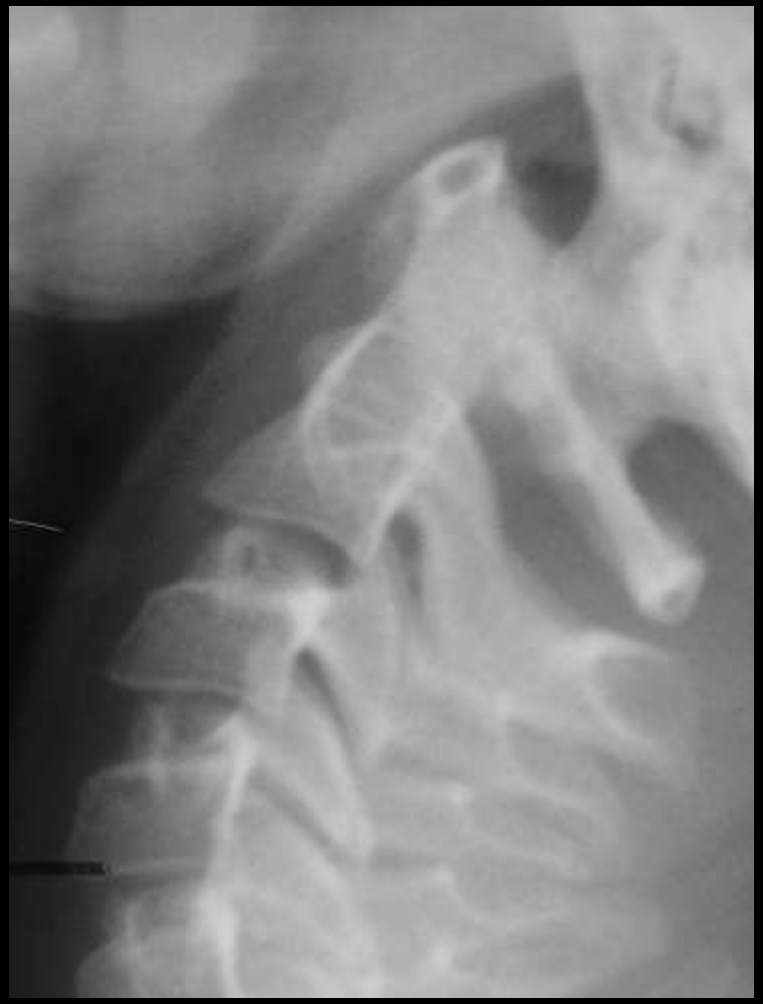
Down's syndrome
Radiology – Hands

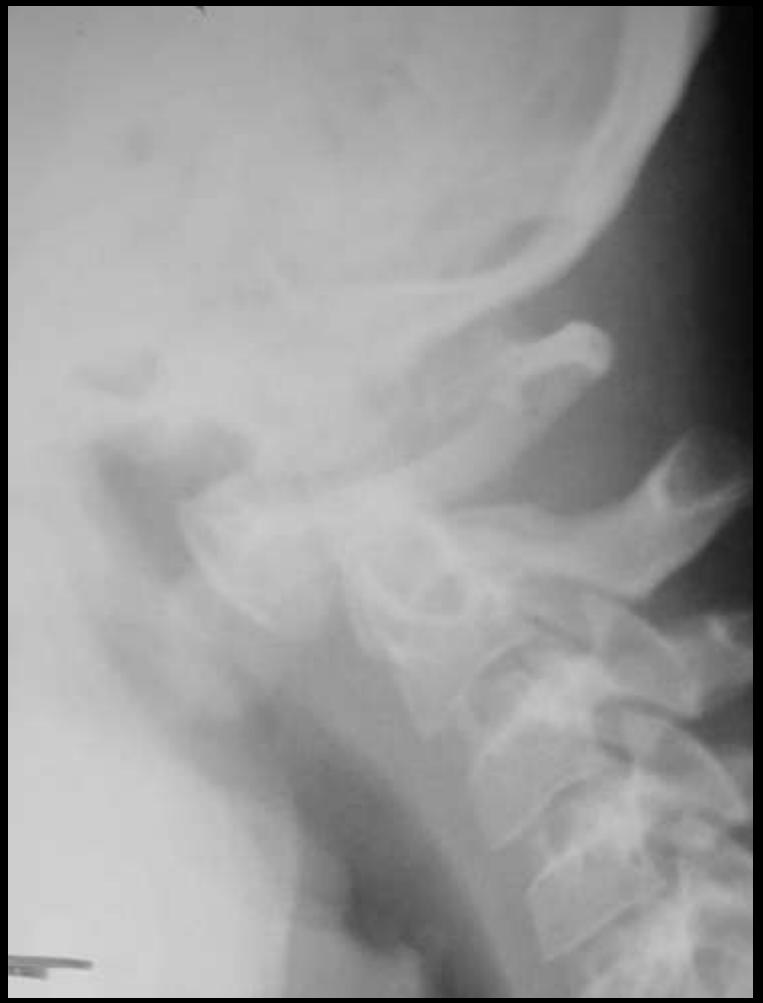
- Short tubular bones
- Clinodactyly (50%)
- Hypoplasia of middle phalanx of little finger (60%)

Turners Syndrome
General

- Females with XO chromosome pattern
- Small stature with retarded bone maturation
- Mental retardation in 10%
- Osteoporosis

Turners Syndrome





Turners Syndrome
Radiology – Axial skeleton

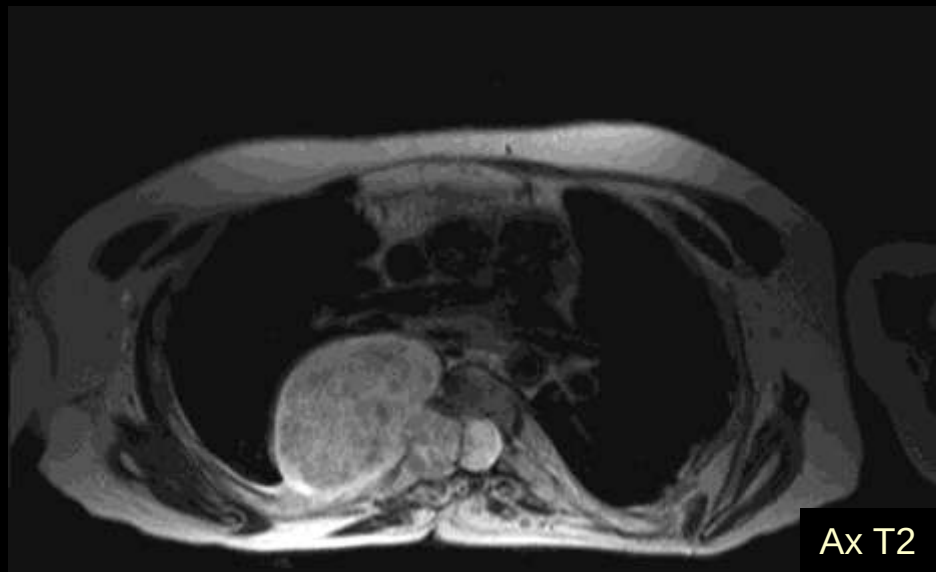
- Pectus excavatum
- Scolosis and kyphosis
- Hypoplasia of cervical spine

Turners Syndrome
Radiology – Appendicular skeleton

- Cubitus valgus 70%
- Short 4th MC +/- MT 50% +/- short 3rd and 5th
- Madelungs deformity
- Enlarged medial tibial plateau +/- small exostosis
- Pes cavus

Neurofibromatosis

- Von Recklinghausen's disease of bone 1882
- Phakomatosis – neurocutaneous syndrome
- 8 variants - NF I-VIII
- 90% NF -1 9% NF -2
- Autosomal dominant, NF1-Cr. 17, 50% mutations
- Mesodermal dysplasia



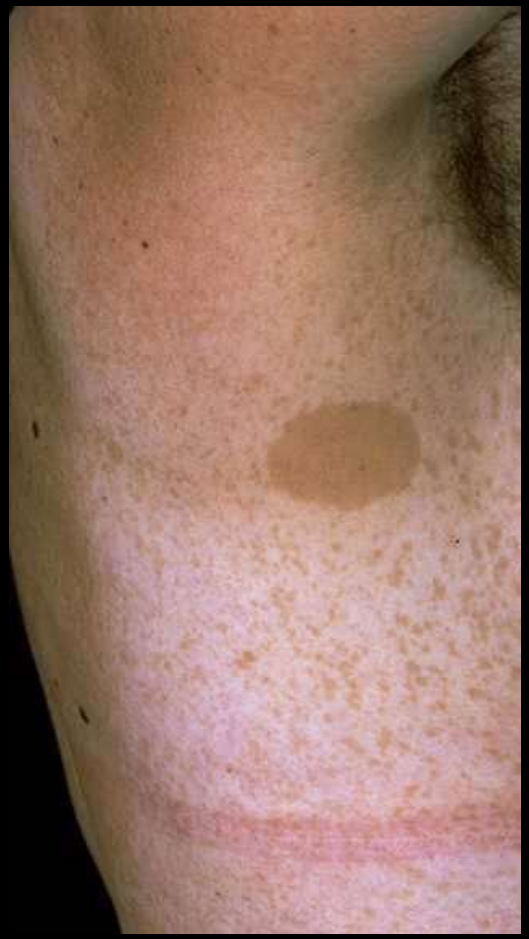
Neurofibromatosis
Clinical triad

- Cutaneous lesions
- Skeletal deformity
- Mental retardation

Neurofibromatosis - Criteria

- 6 or more café-au-lait
  - >5mm prepubertal
  - >15mm postpubertal
- >1 neurofibromas or one plexiform
- Axillary or inguinal freckling
- Optic glioma
- >1 Lisch nodules
- Distinctive bone lesion
  - Sphenoid dysplasia
  - Pseudo arthrosis
- 1st degree relative

- 2 or more of the above



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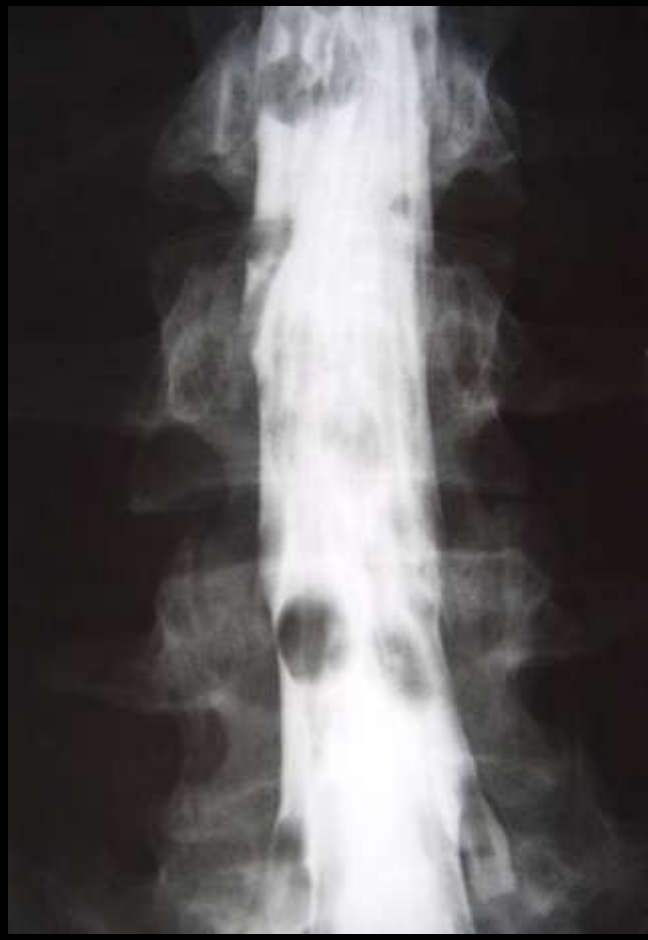

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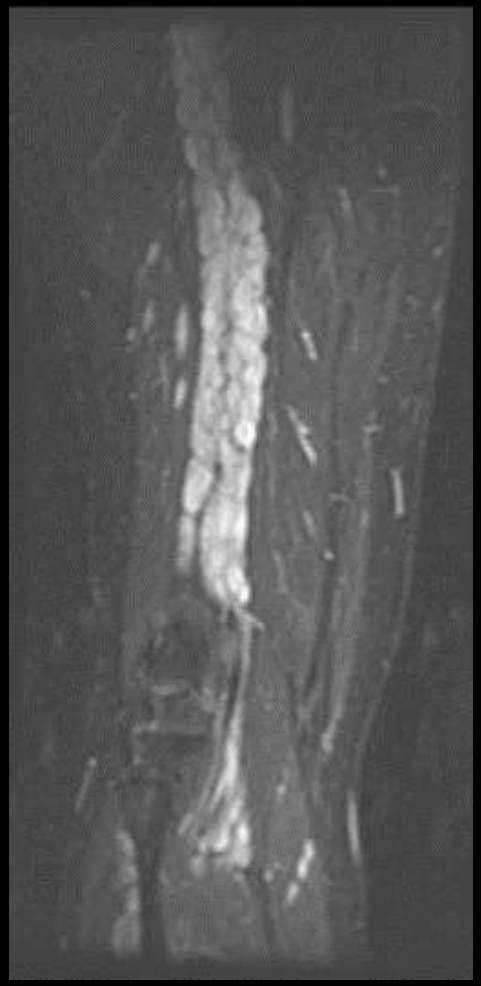

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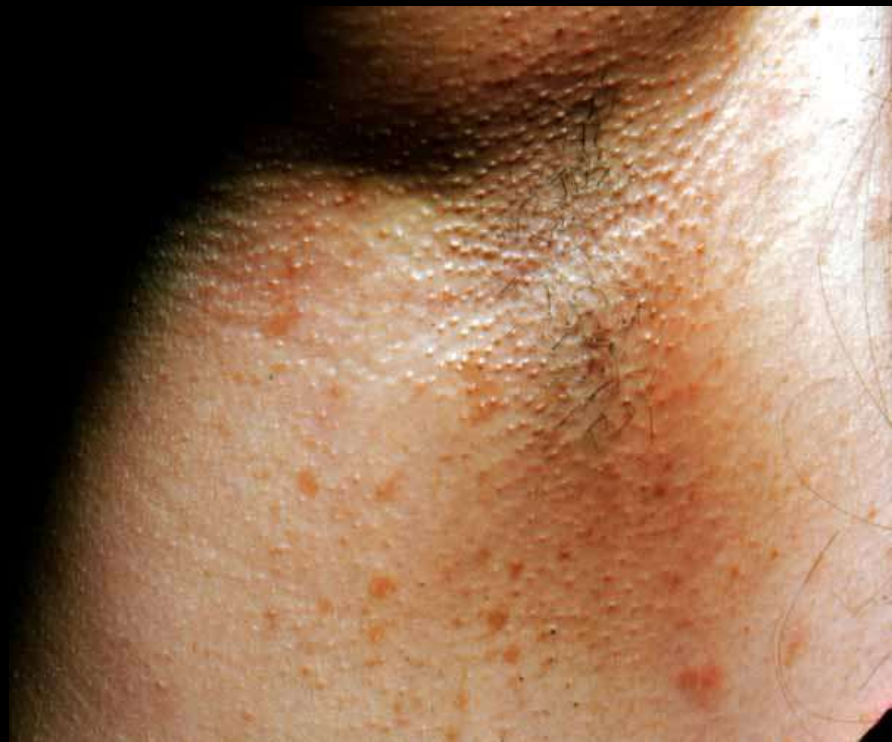

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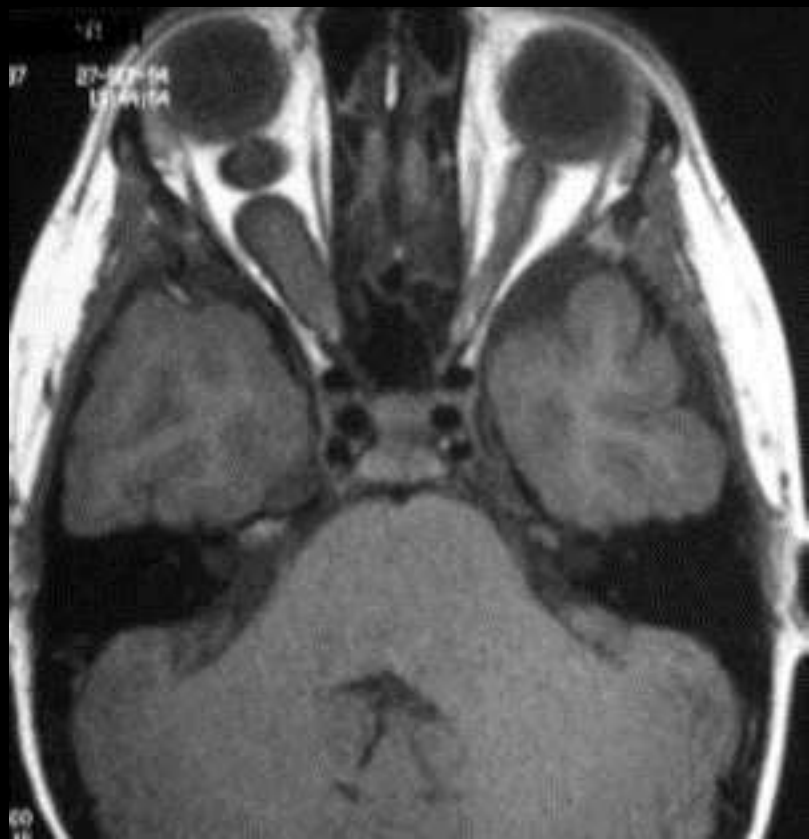

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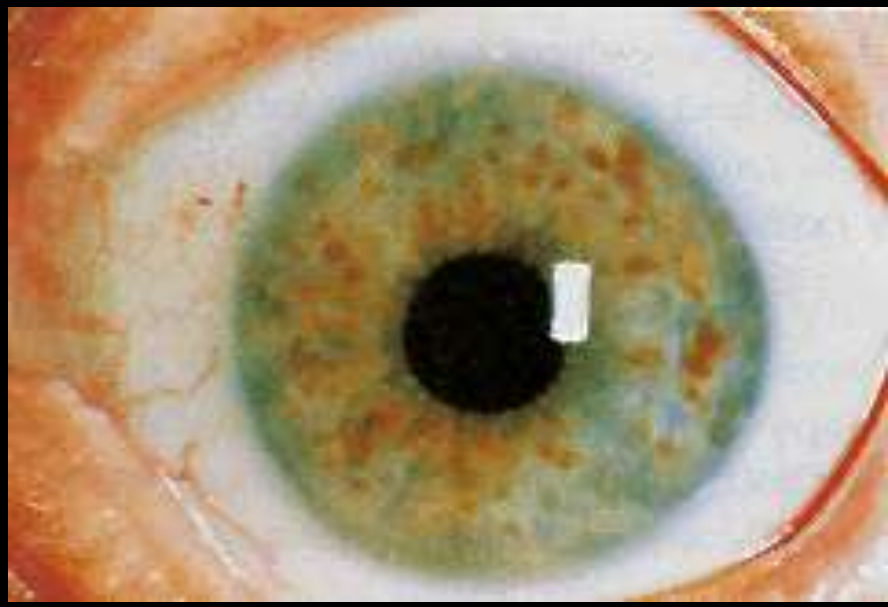

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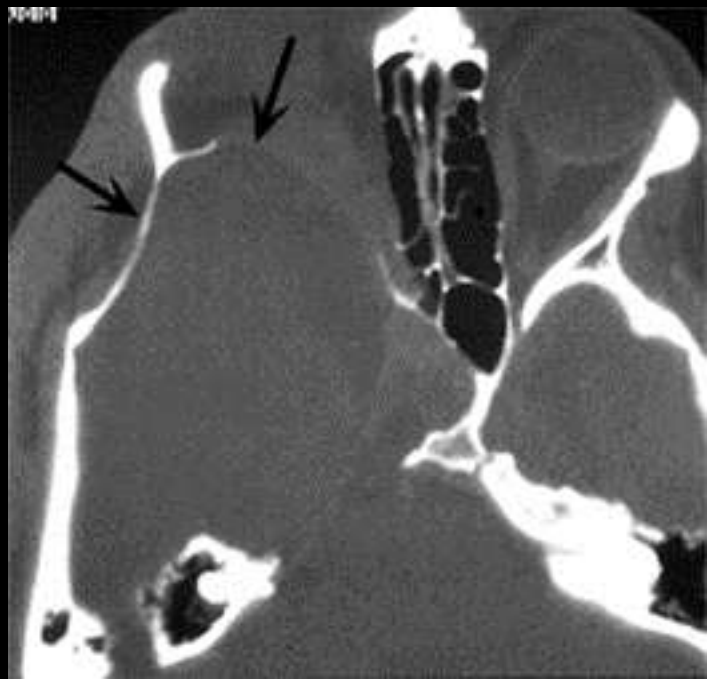

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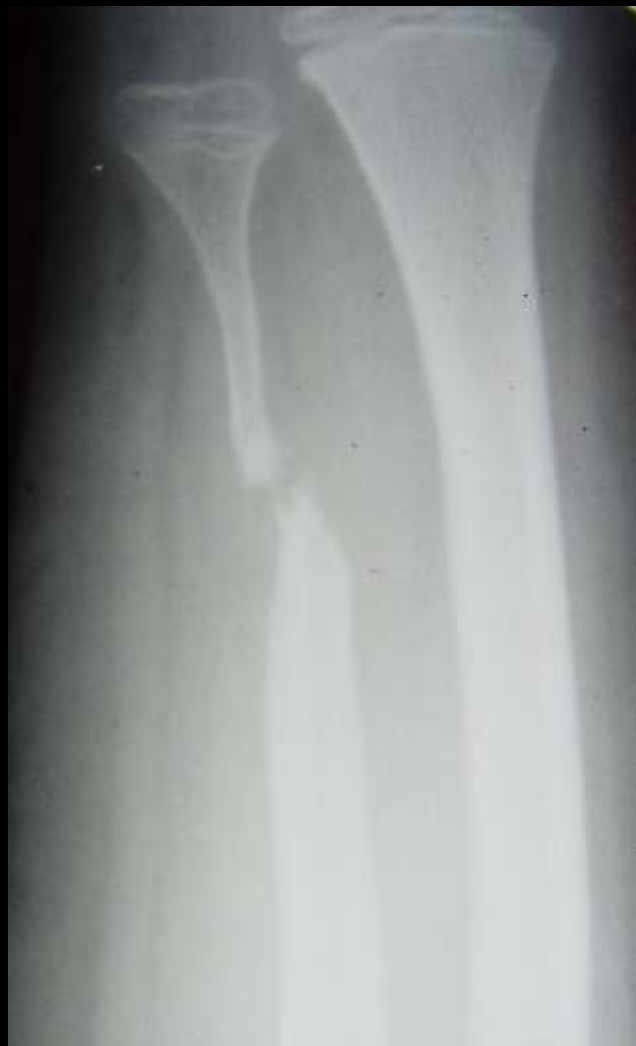
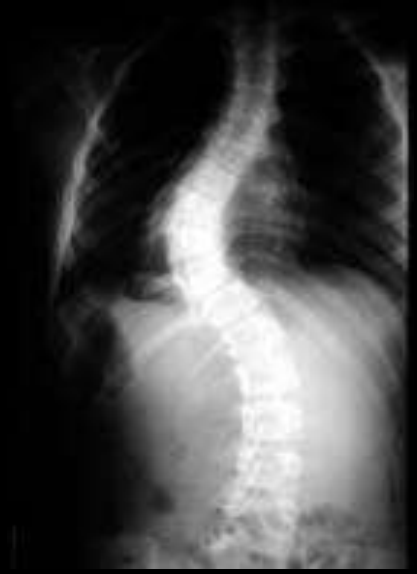


Osseous abnormalities of NF1

- Scoliosis (short or long segment)
- Kyphosis (often predominates)
- Facial or orbital dysplasia
- Lambdoid suture defects (left sided)
- Pseudoarthrosis (tibia + congenital)
- Periosteal abnormalities (reaction or cyst)
- Multiple NOF or fibroxanthomas
- Rib deformity (ribbon ribs)
- Posterior vertebral scalloping (dural ectasia)
- Elephantiasis neuromatosa
- Mesodermal dysplasia pseudofracture



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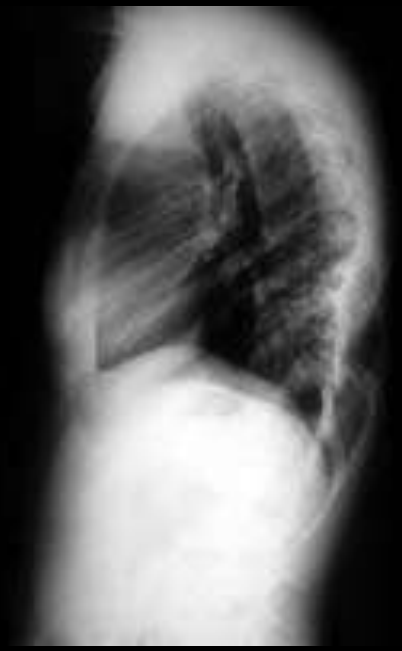

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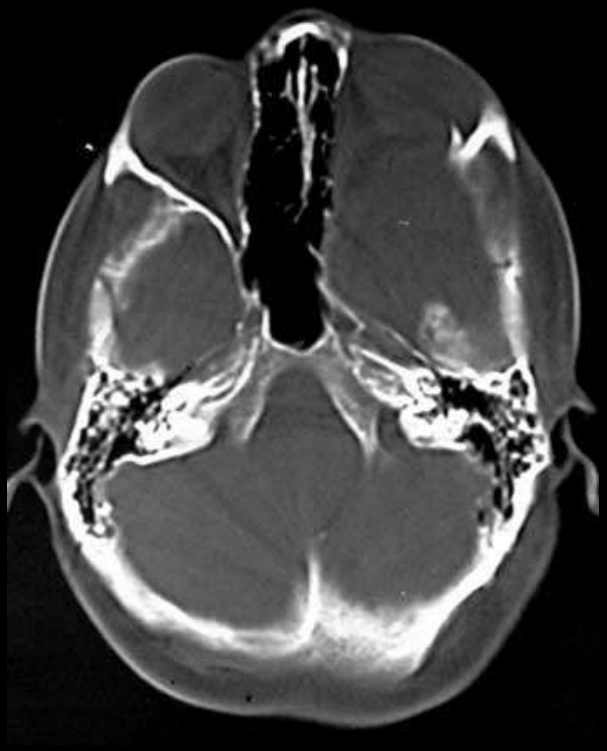

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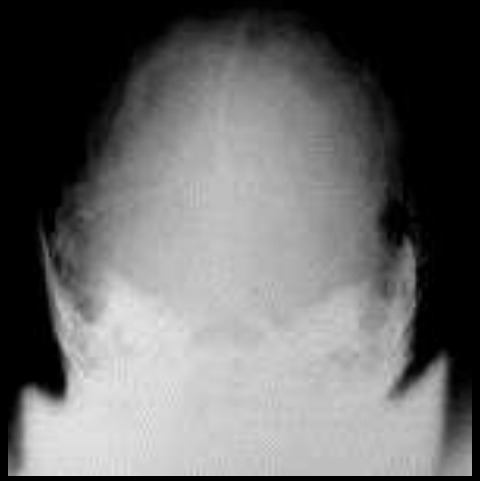

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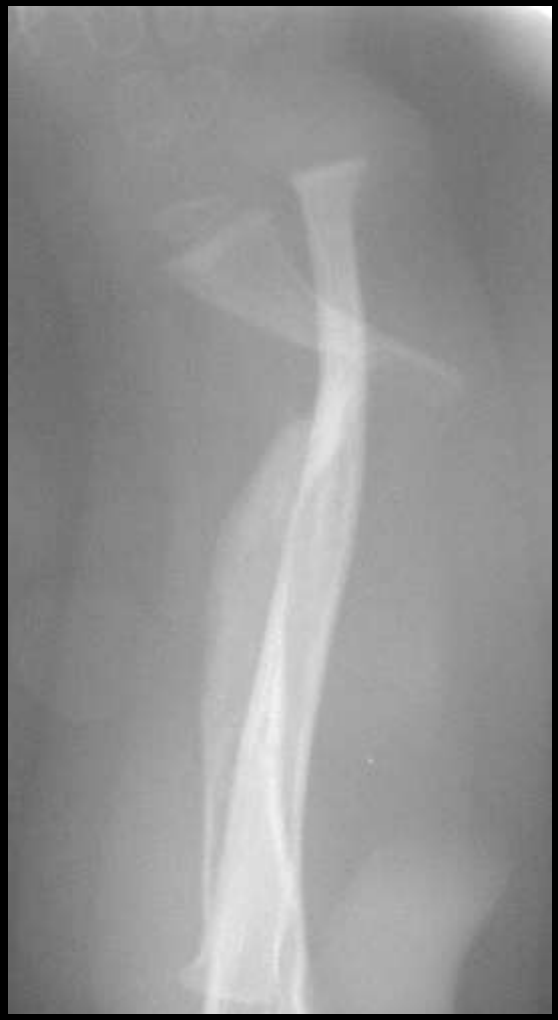

Nerve Tumors - Neurofibromatosis

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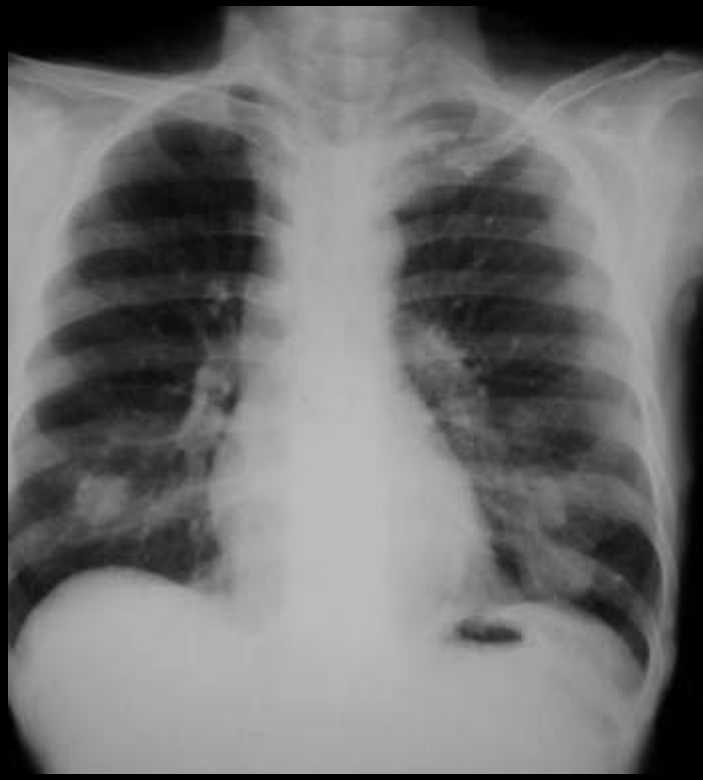

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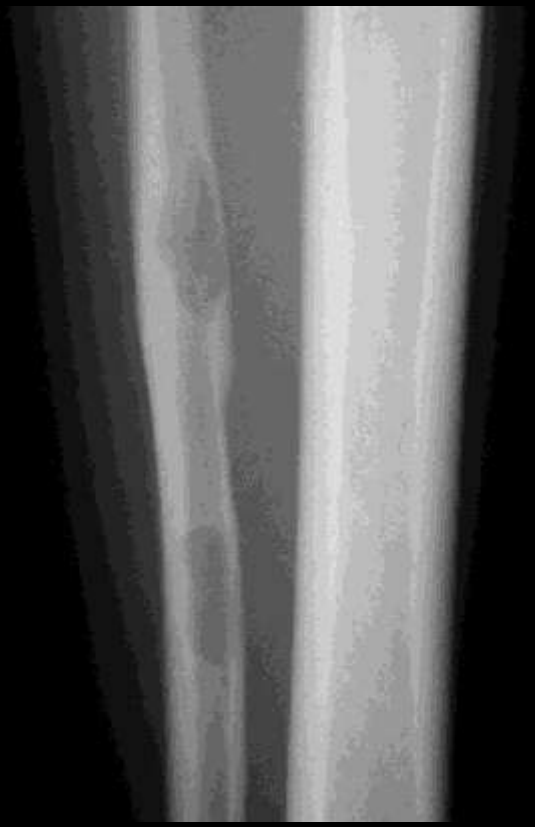

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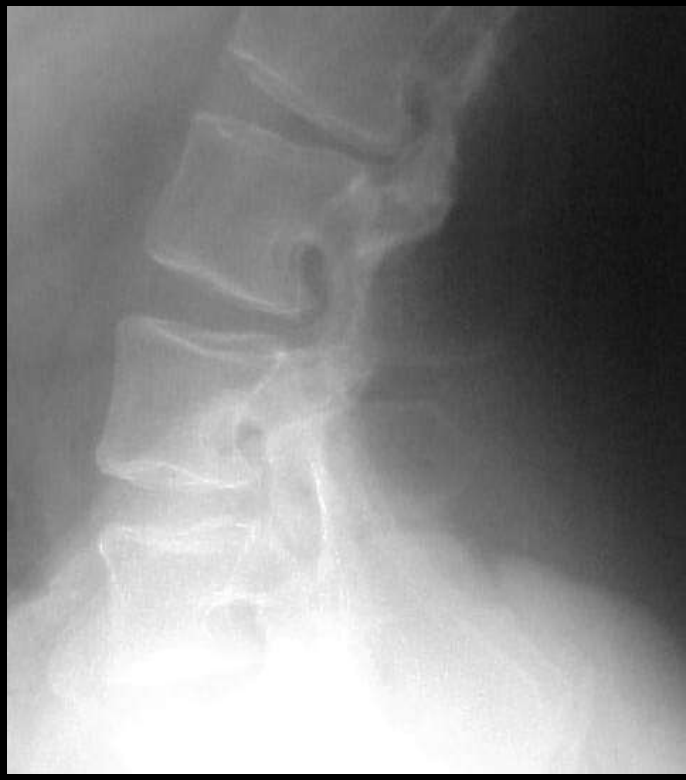

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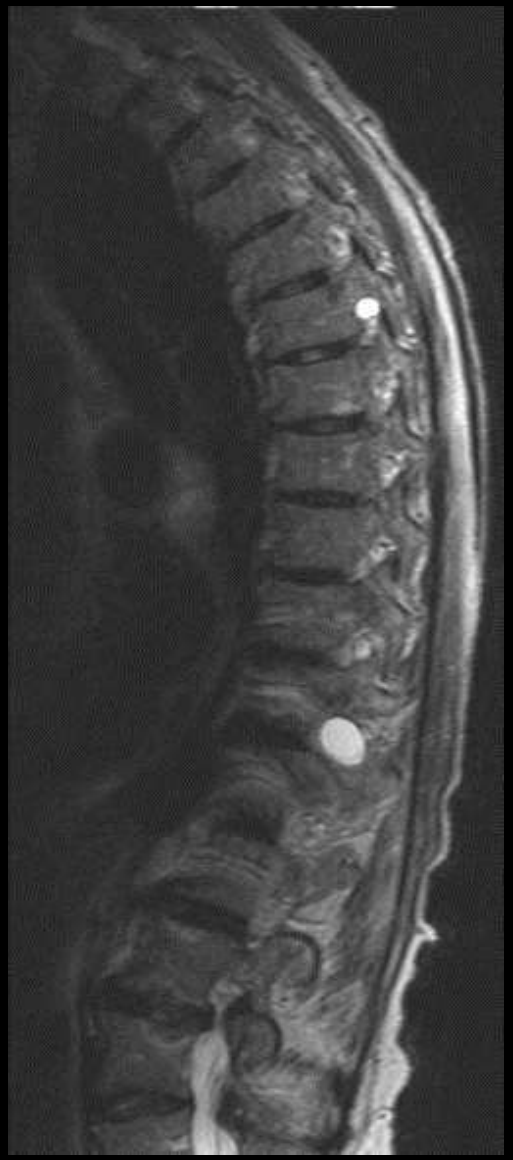

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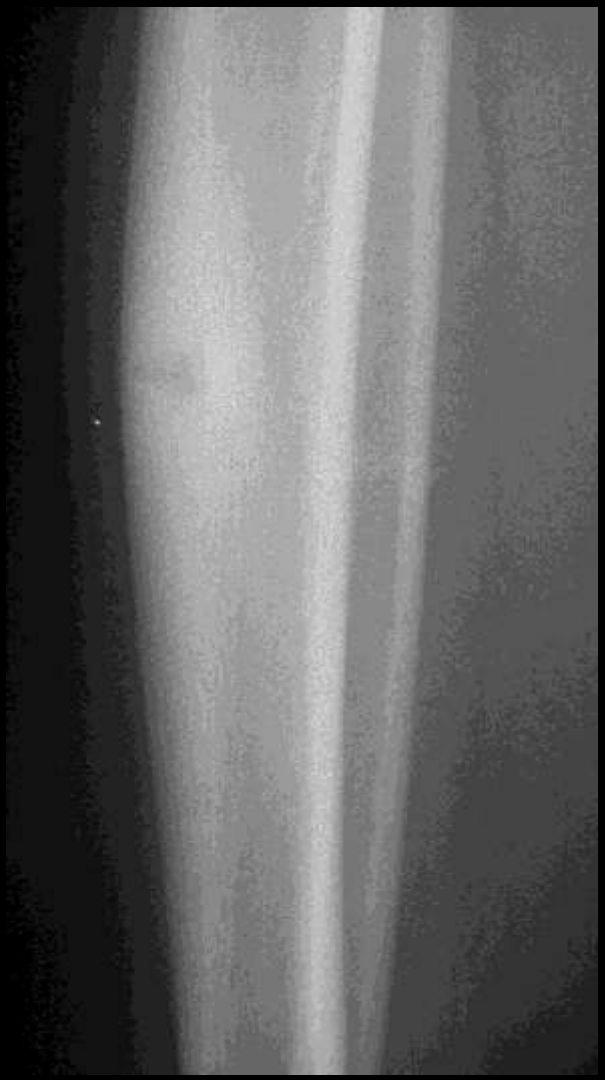
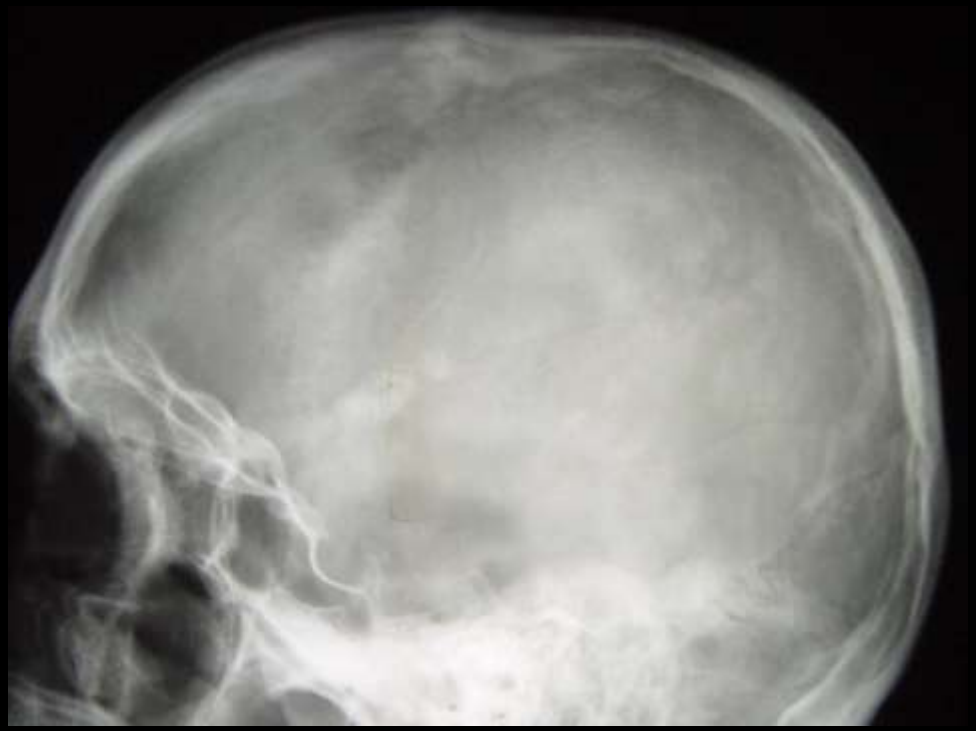
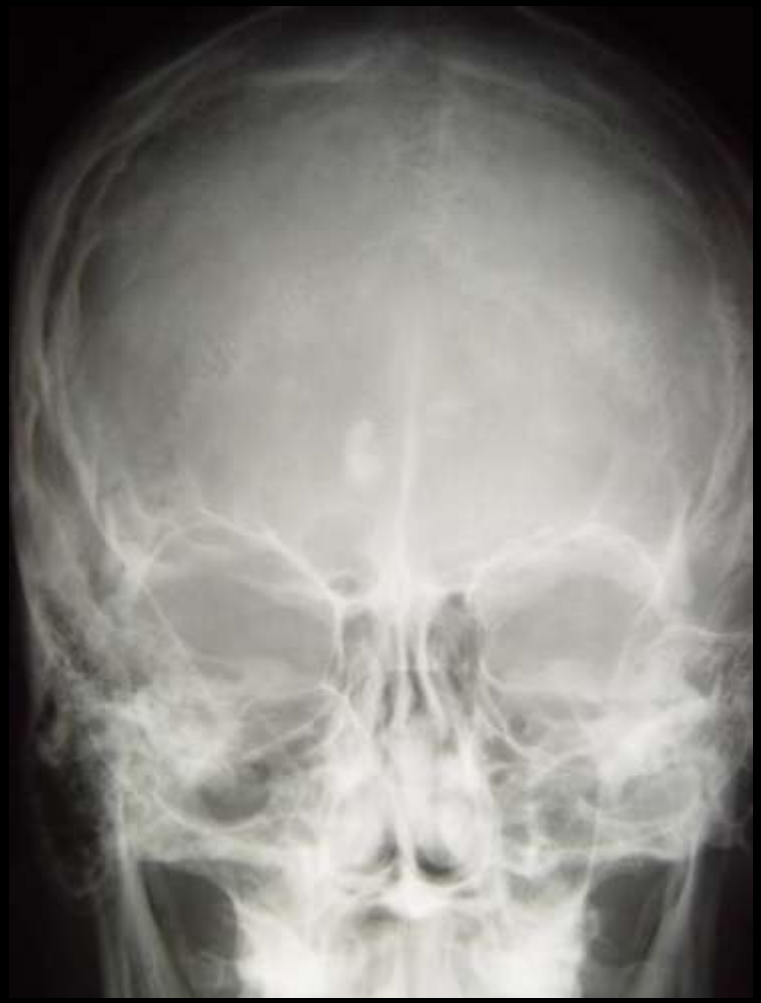


Tuberous Sclerosis





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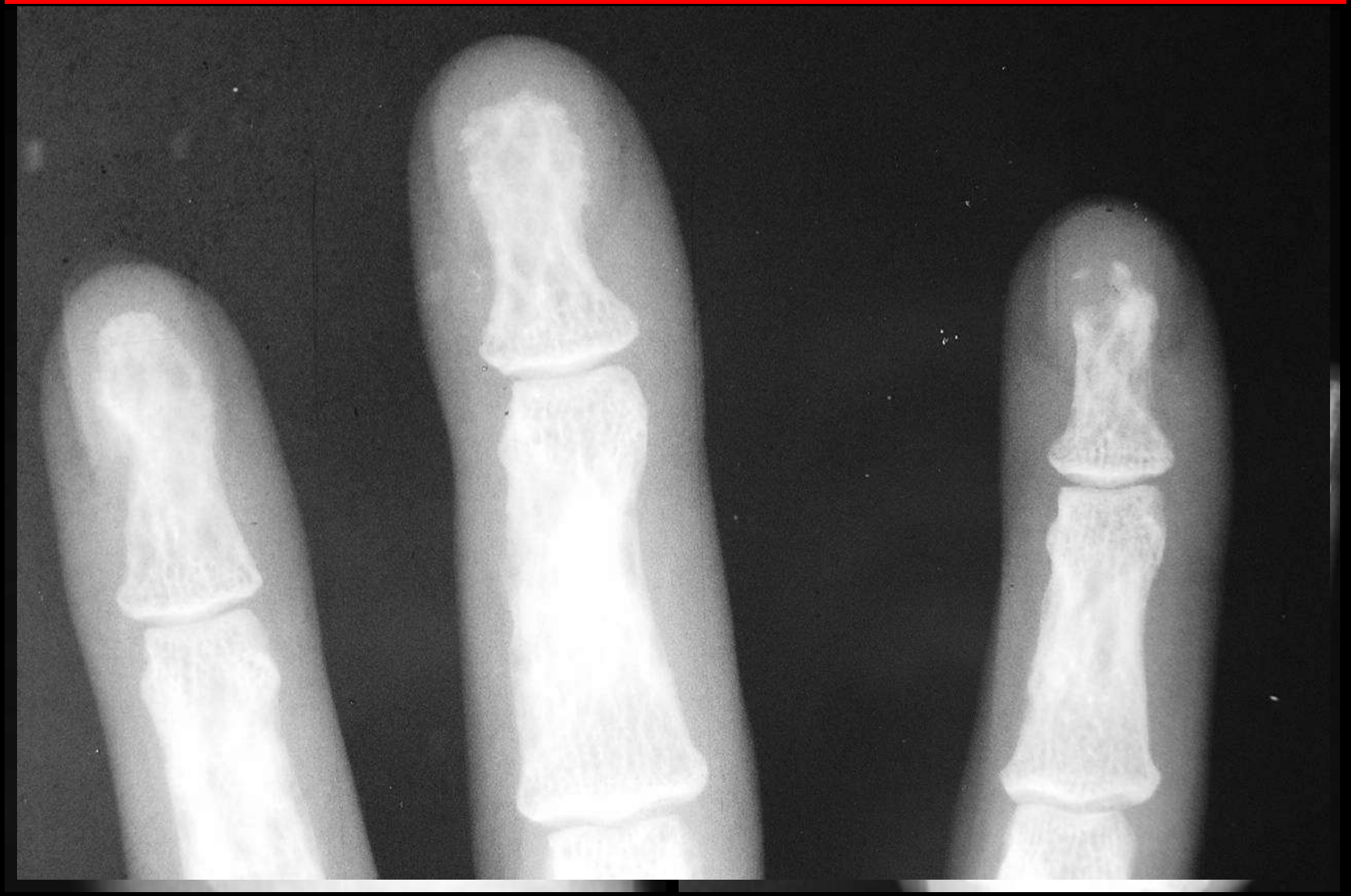


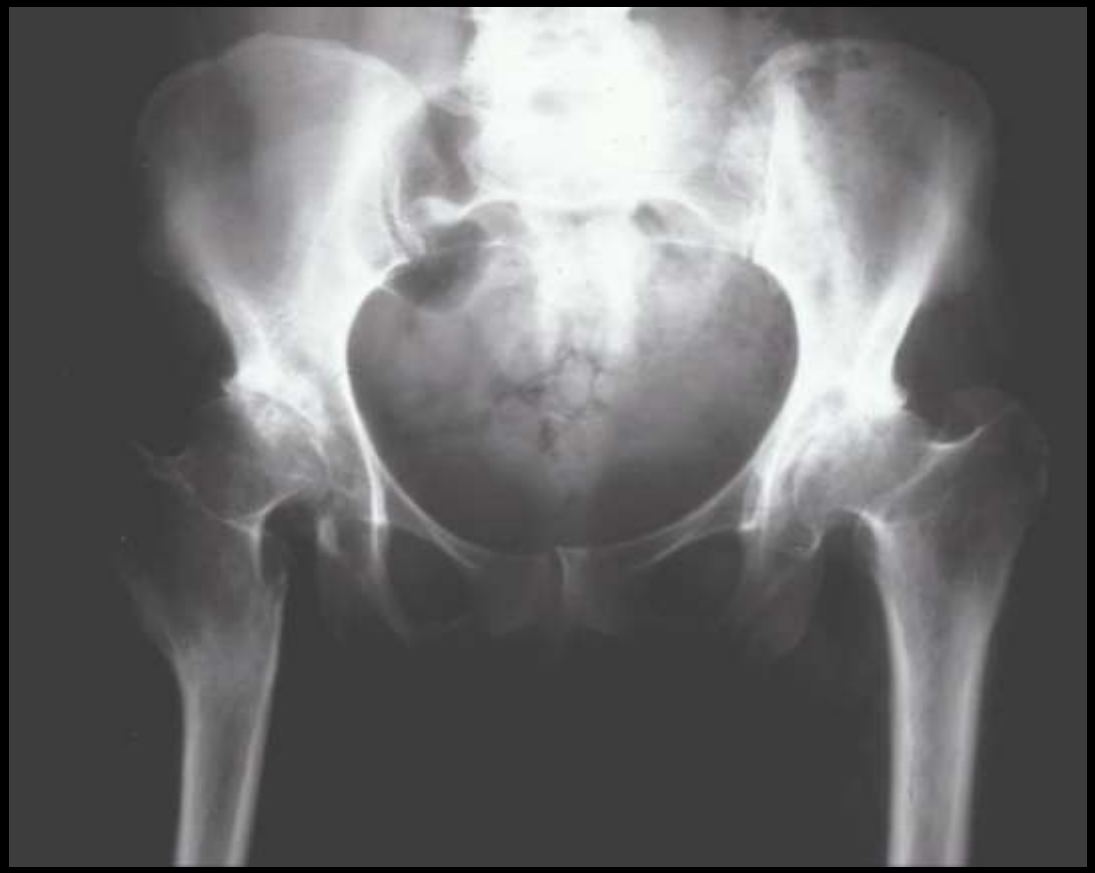
Tuberous Sclerosis
General

- AD, 25-50% fresh mutations
- Mental retardation 60%
- 75% dead by 20 years

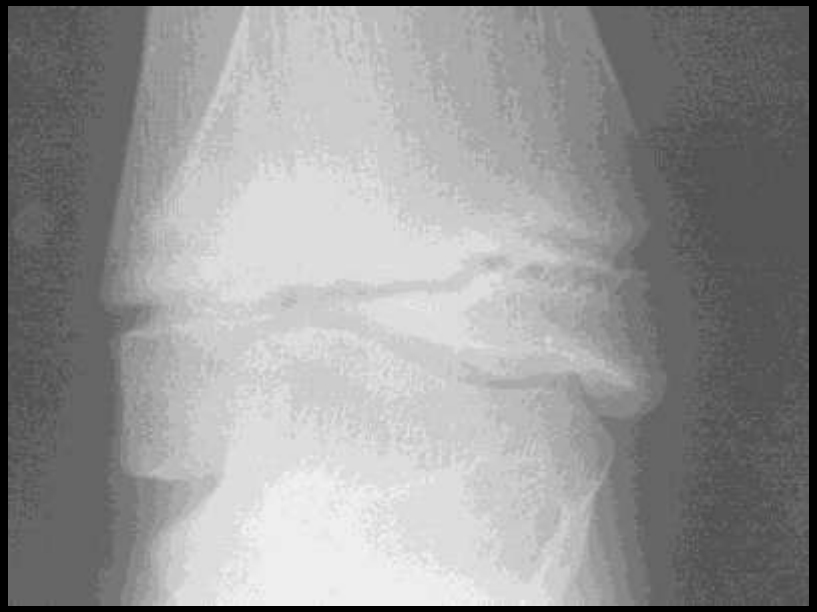
Tuberous Sclerosis
Radiology

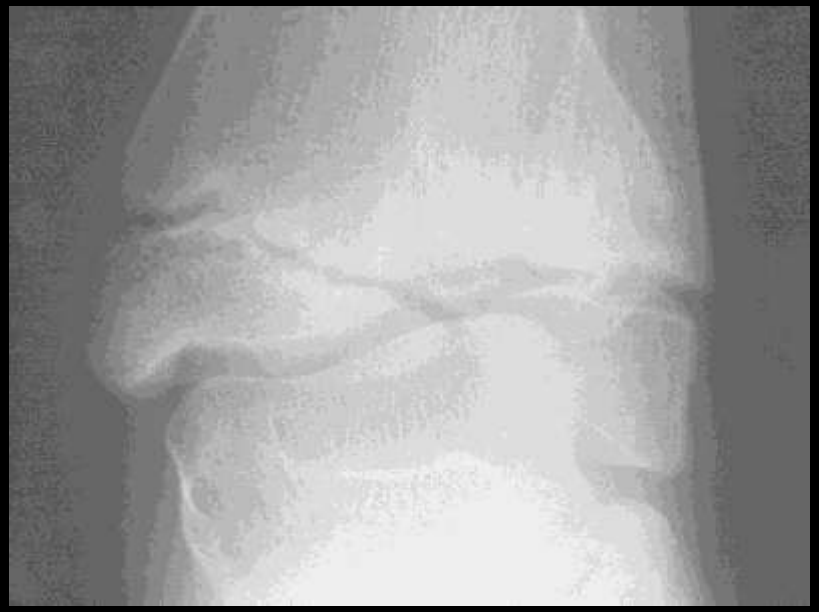
- Sclerotic islands in 50%
  - Calvarium, spine, pelvis
- Hands > Feet
  - Cystic defects
  - Periosteal new bone

Spondyloepiphyseal Dysplasia



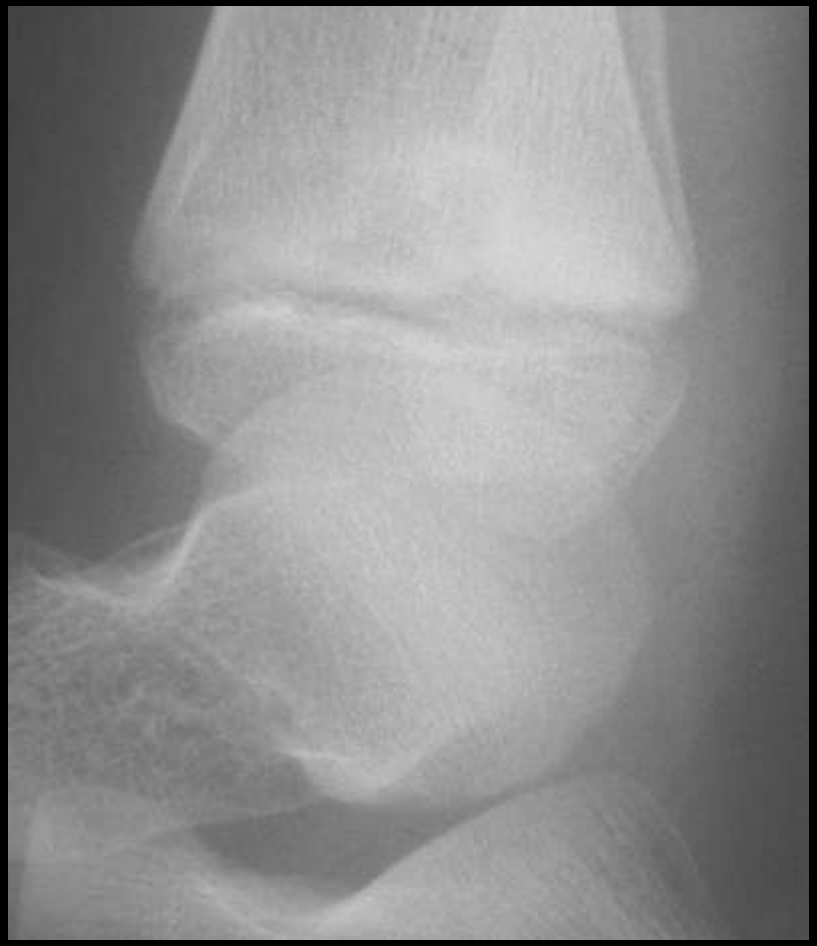
Multiple Epiphyseal Dysplasia

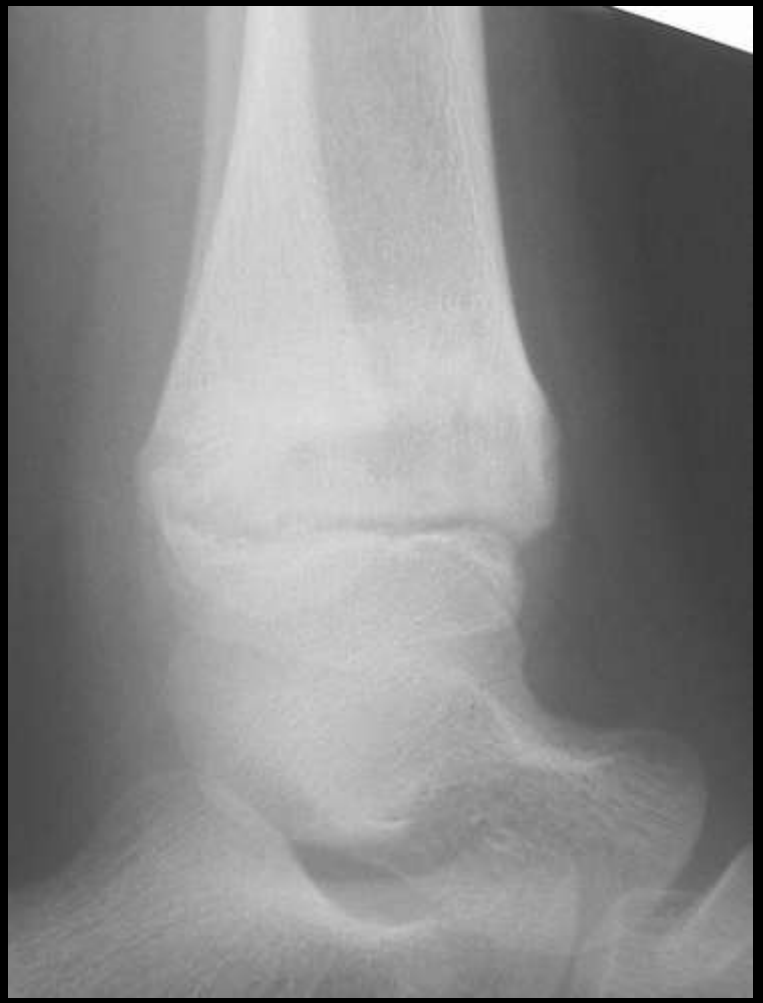




12M

Multiple Epiphyseal Dysplasia





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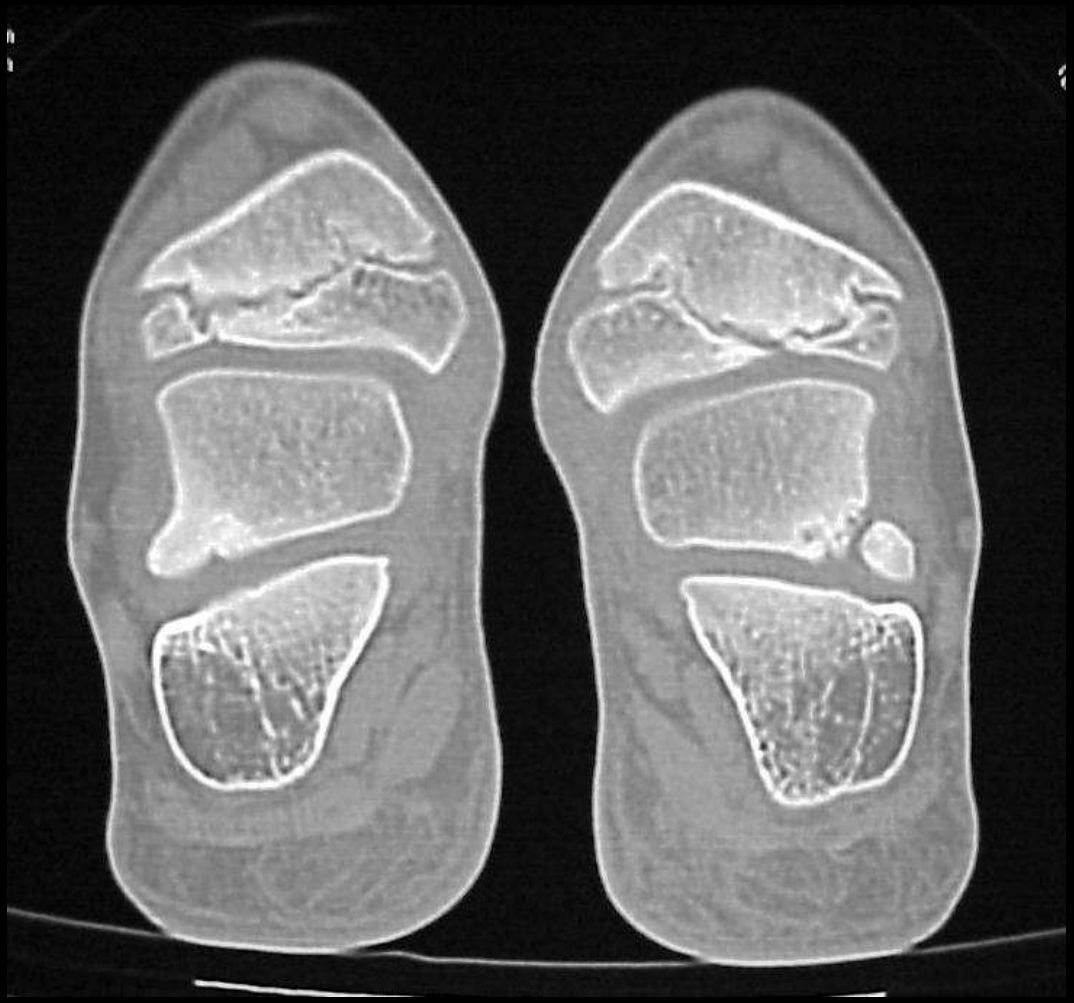

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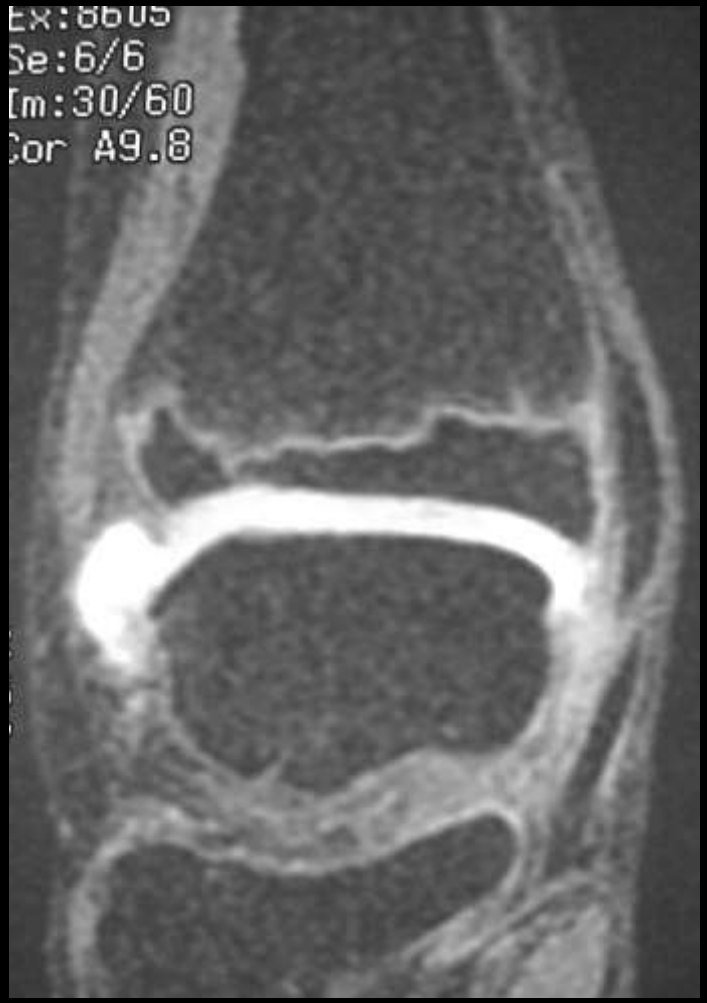

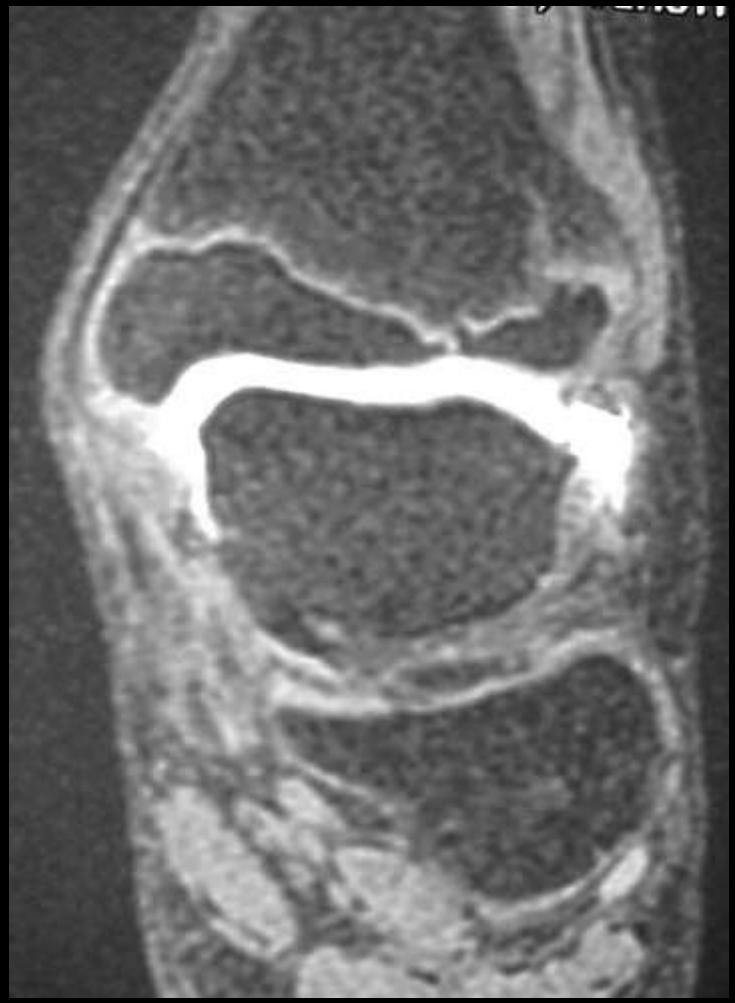

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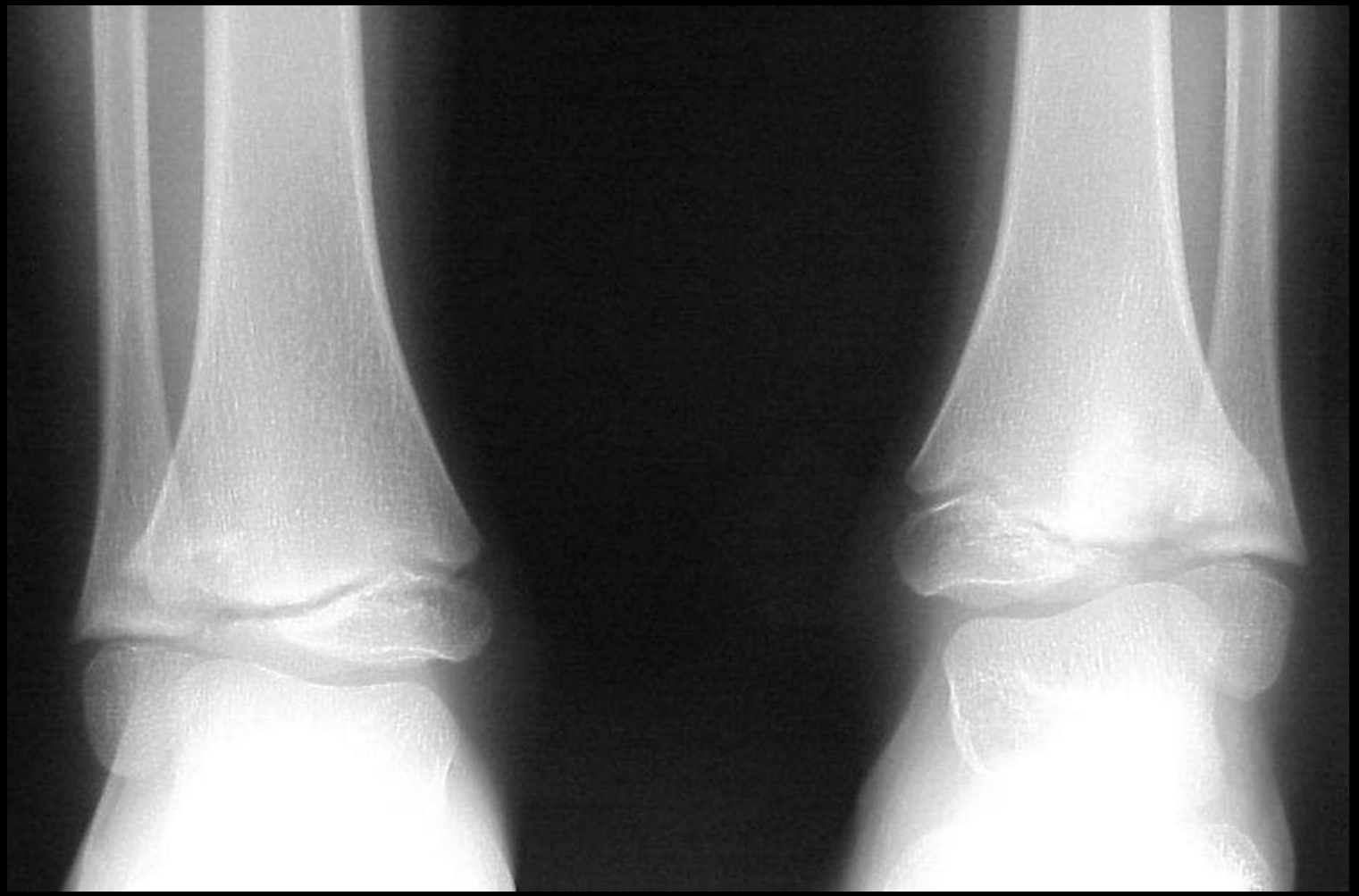

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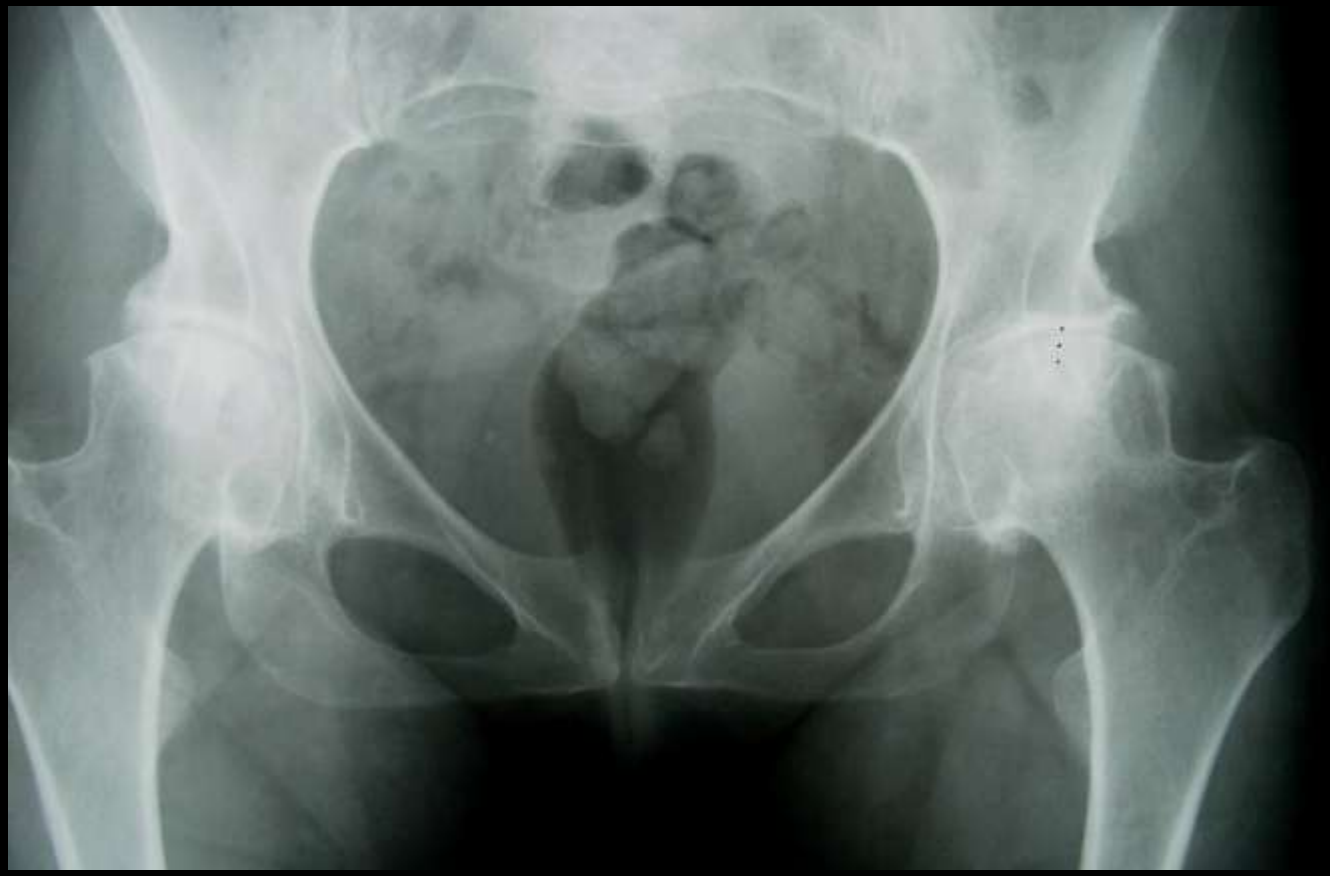

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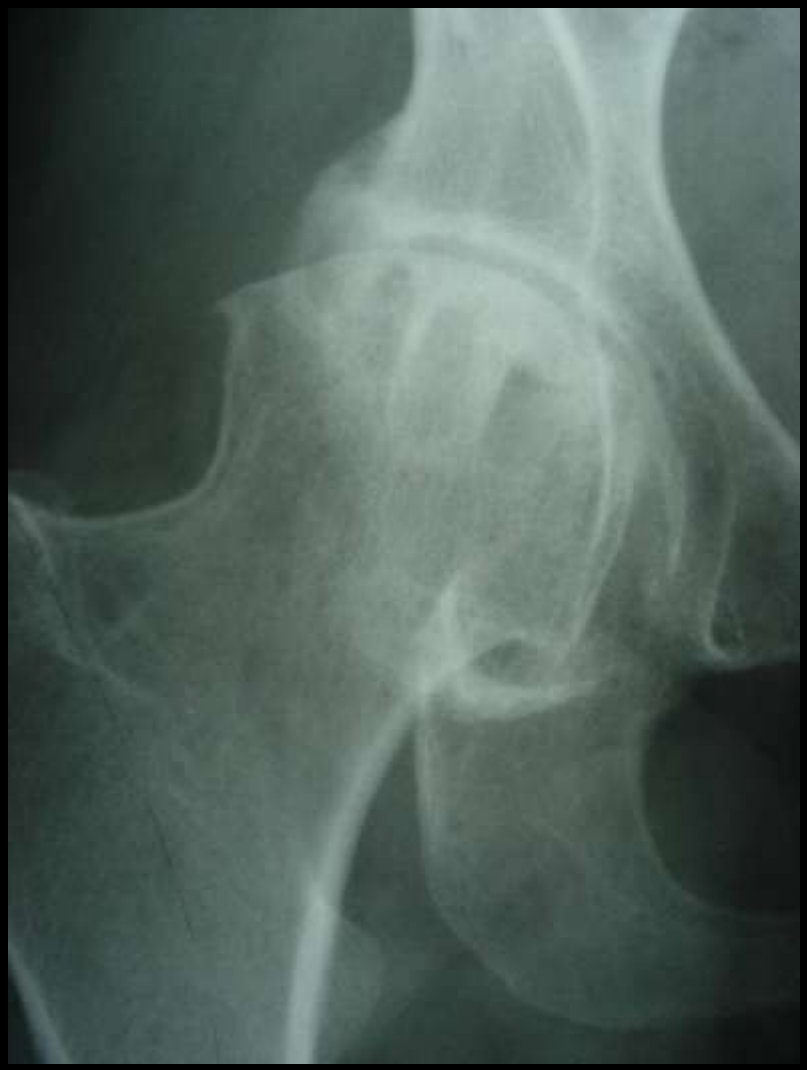
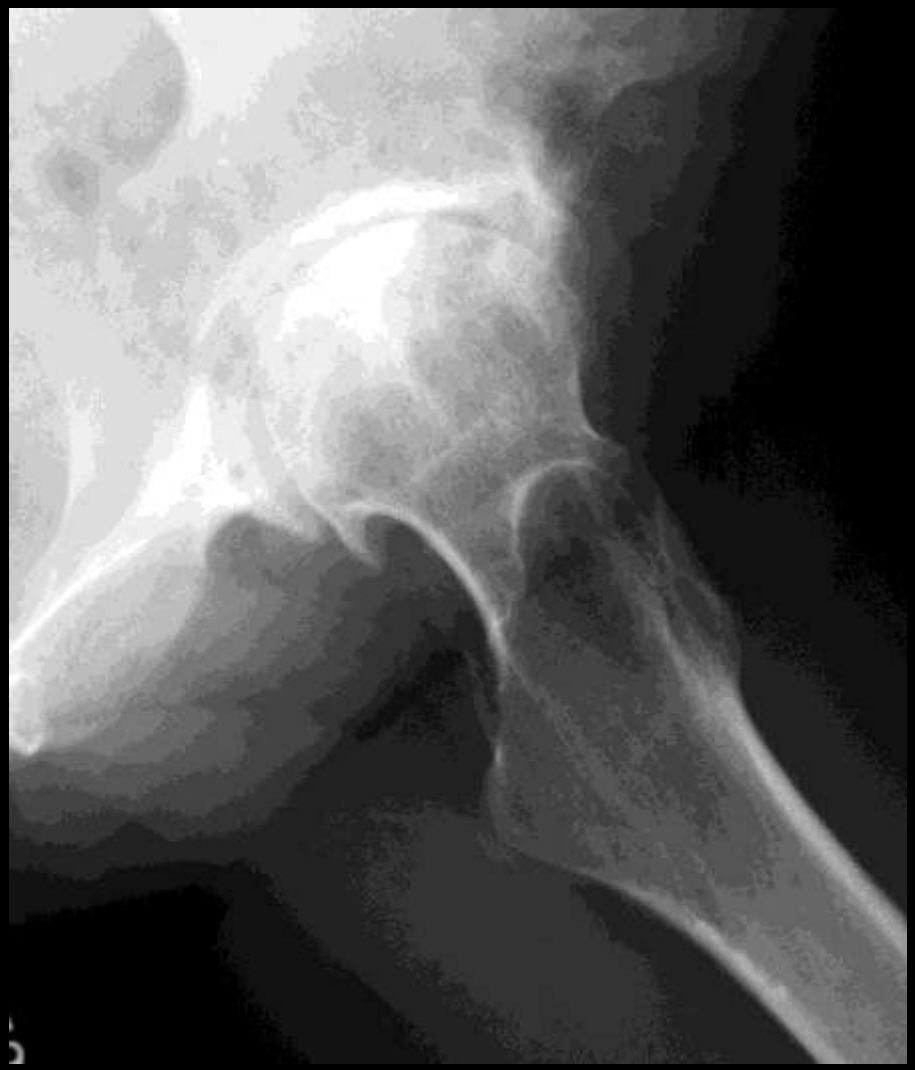
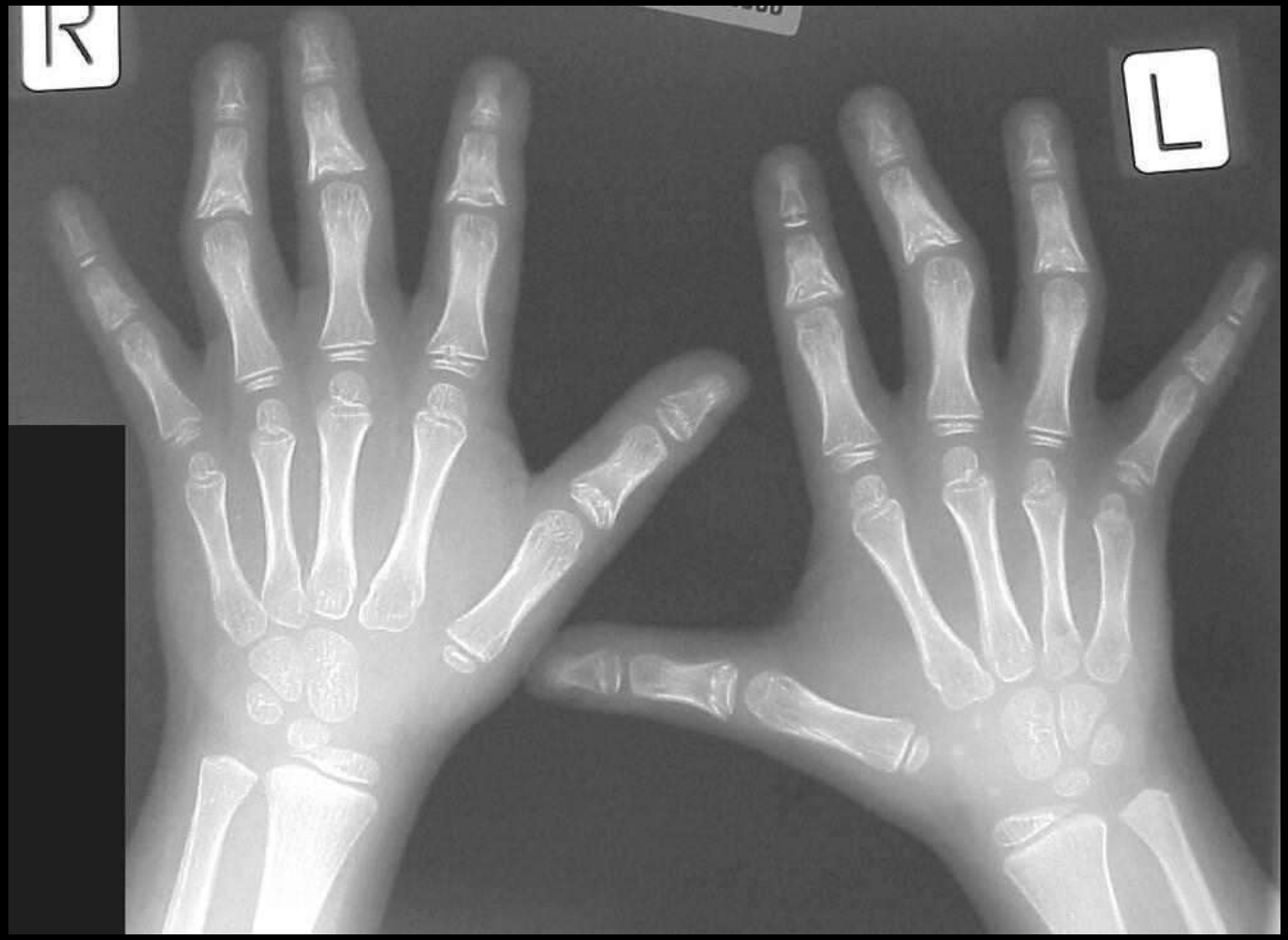




Trichorhinophalangeal Syndrome of Giedion



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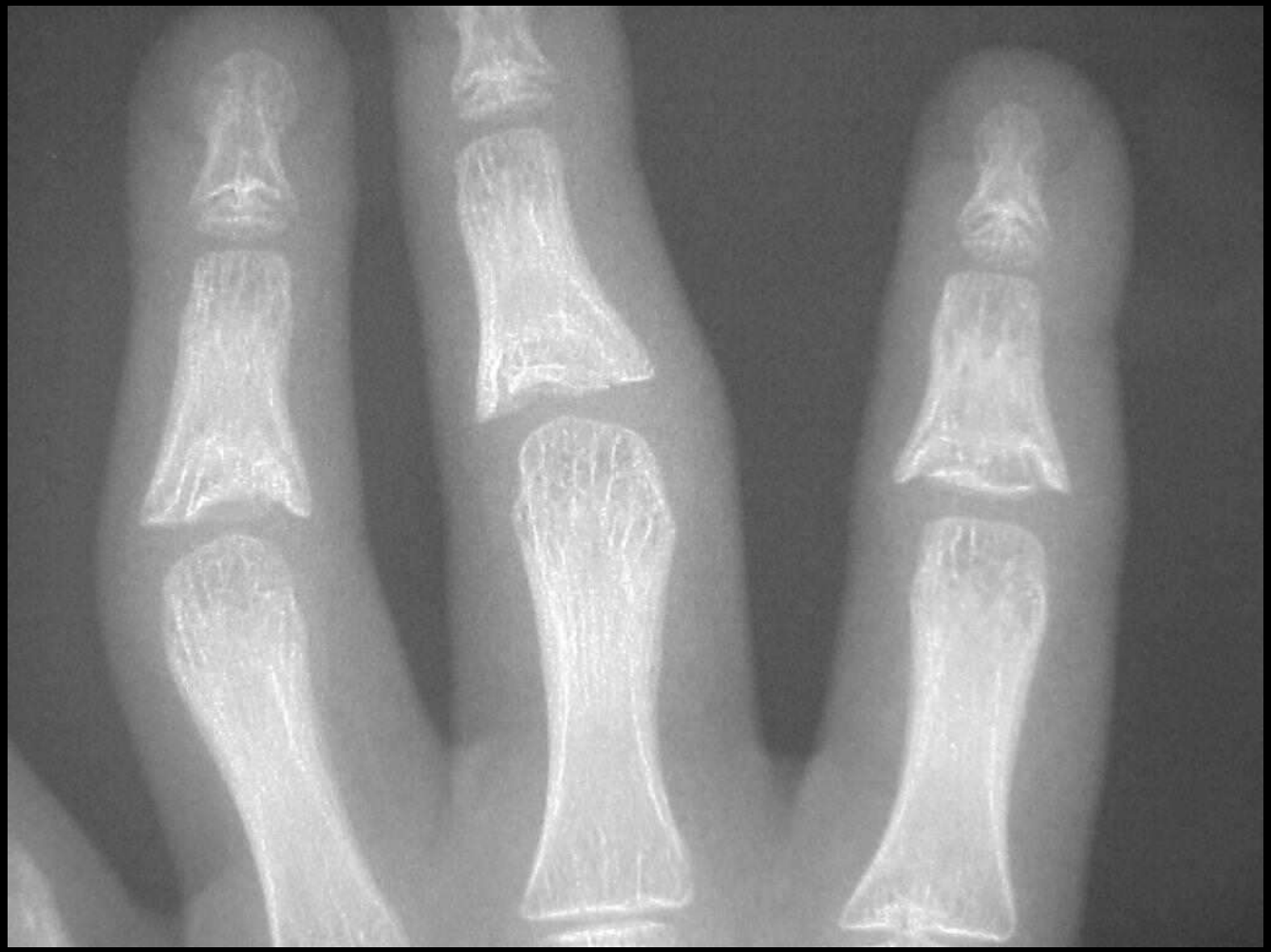
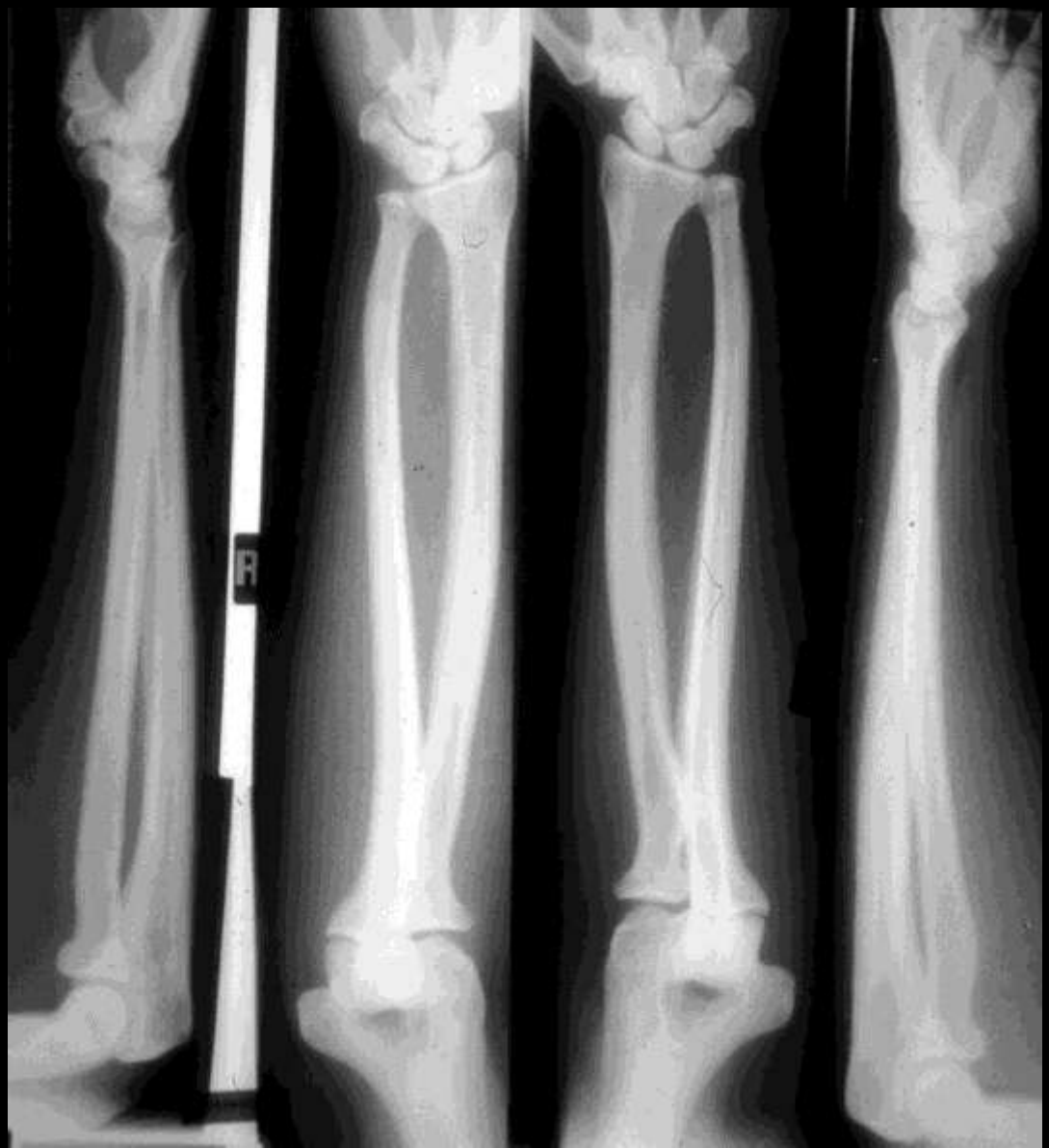
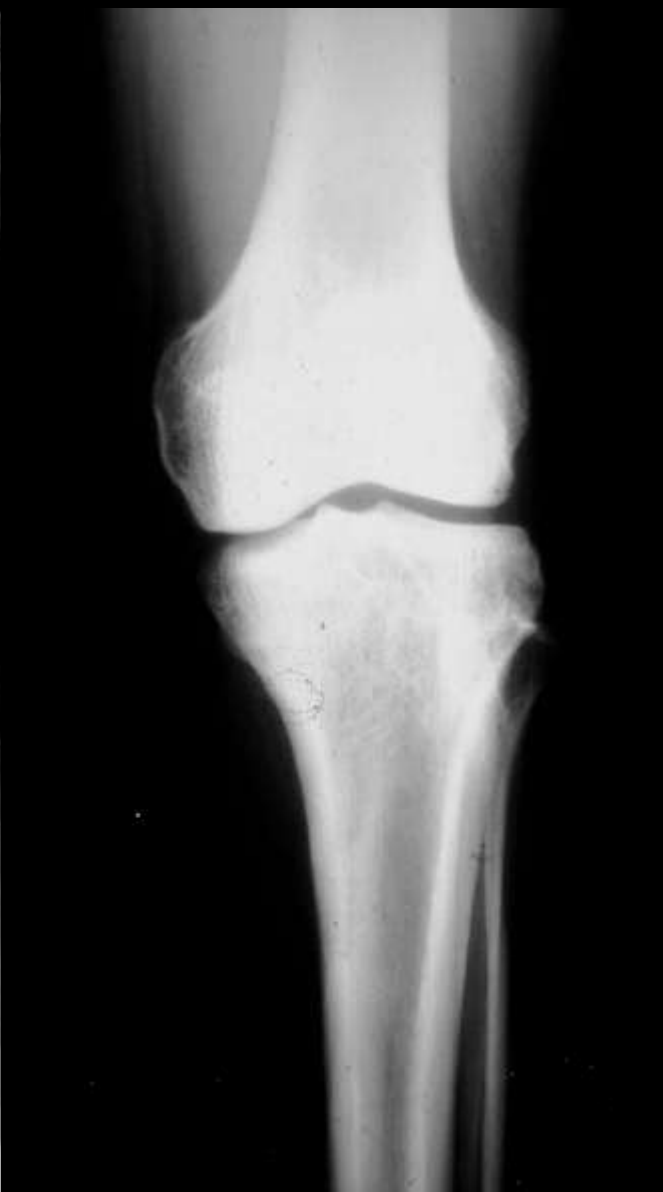


Camurati Engleman





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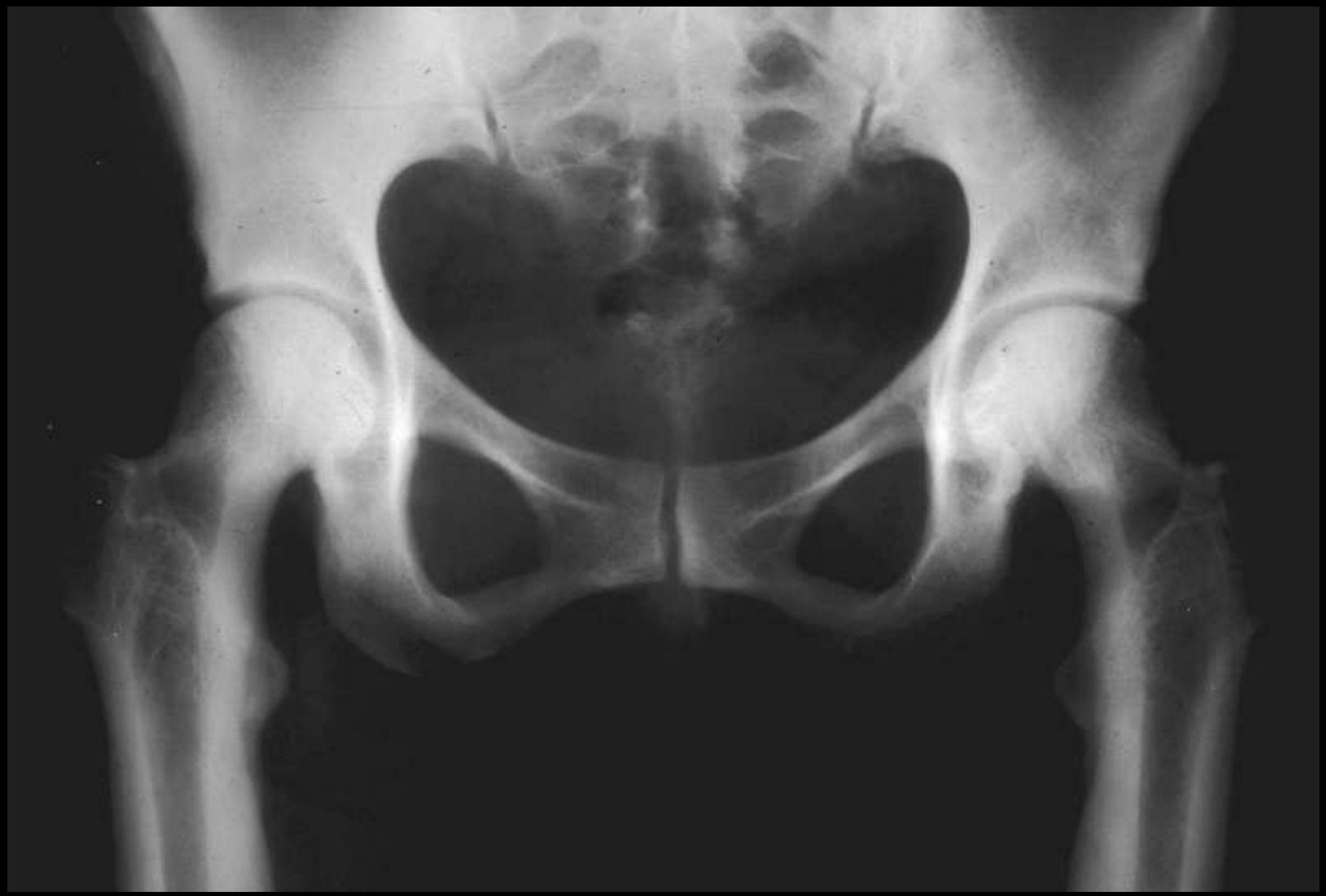

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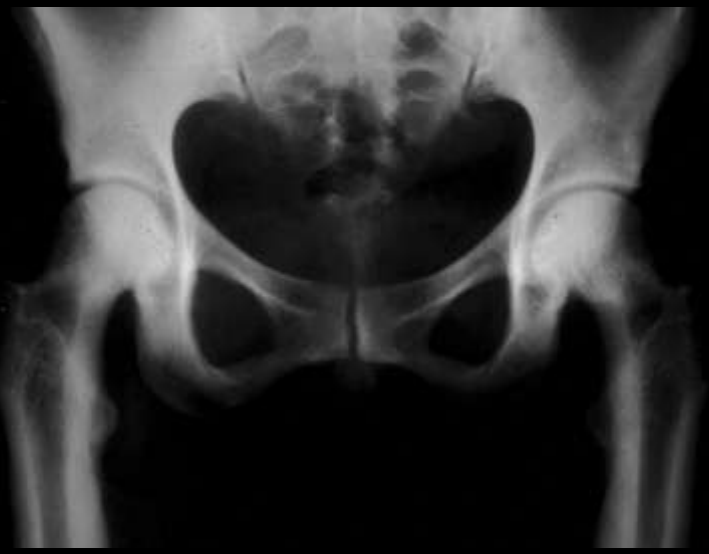

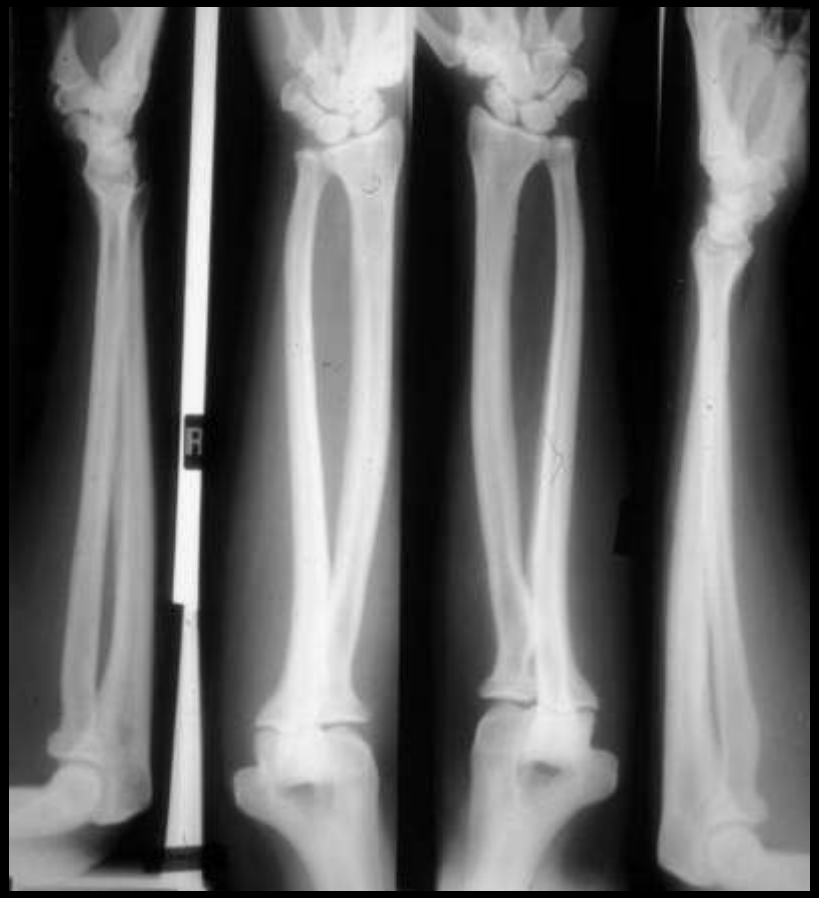

Ribbings disease