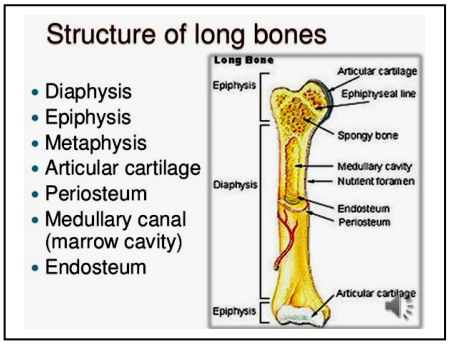
SKELETAL DYSPLASIA

- **Def.**: Generalized abnormalities in bone growth & development.

  ⇨ **Etiology :** - Hereditary - Gene Mutation

  ⇨ **Classification** : Large variable types of diseases, These are most famous ,

  ⇨ ***Disorders affecting:***

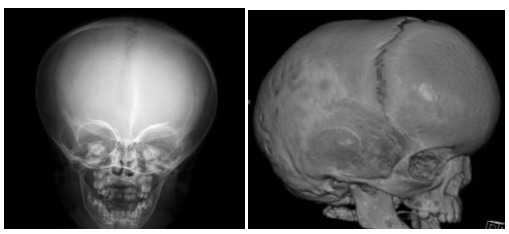


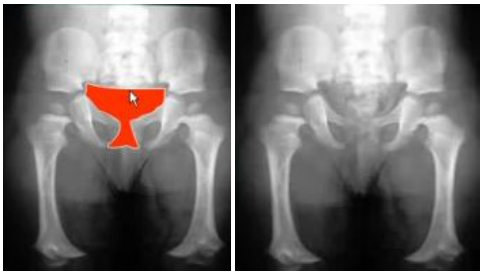
1-Growth plate	2-Epiphysis/Apophysis	3-Metaphysis	4-Dyaphysis	5-Others
-**Achondroplasia** -HypoChondroplasia -Psudo-Achondroplasia -*Thanato phoric dwarfism "rare, died IU"*	-**Multiple** Epiphyseal Dysp. -**Spondylo** " " -**Punctate** " " -**Hemimelica** " " تحدث فى نصف الجسم وتترك النصف الاخر - …… & ***Others***	-Chondrodysplasia -Osteopetrosis *"Marble Bone disease"* -**Dystosis** : =Cliedo-cranial D. =Pykno D.	-Diaphyseal Dys. -***Pachy-dermo- perisoteosis*** -***Infantile cortical Dysplsia*** -*Hyperphosphatasia* -	**Osteogenesis Imperfect*a *

Achondroplasia "Dwarf"

⇨ **Etiology** : - Autosomal Dominant - 80% Gene Mutation

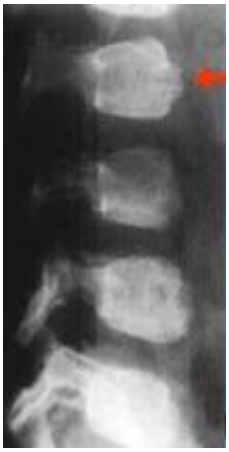
General	-Dwarf: short stature - Short Limbs -Usually Normal mentality
Skull	-**Large** Skull - **Small** (Face – Skull base –Sella) +/- Hydrpcephalus
Chest	-Short broad sternum & Limbs
Spine	⇩Distal interpedicular distance - Ant. Peak -Post Scalloping
Pelvis	-Square iliac bones -Glass shape cavity - Sacral Jutting: displaced post.
Limbs	-Wide metaphysis - Ball & socet end - **R**hizomelia → More short Proximal bones -**Trident Hand**: Equal Finger length "3 equal + other 2 equal in length"

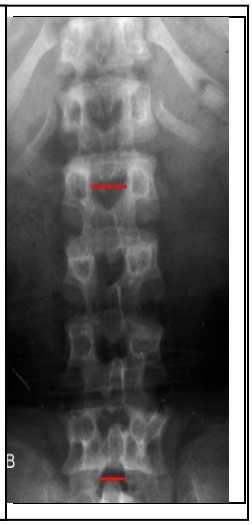




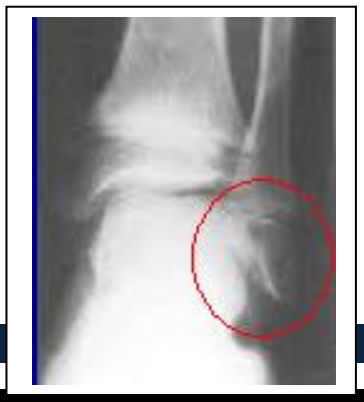
Trident Hand

Ant. Peak





Hypochondroplasia



…….. ***As Achondroplasia*** +

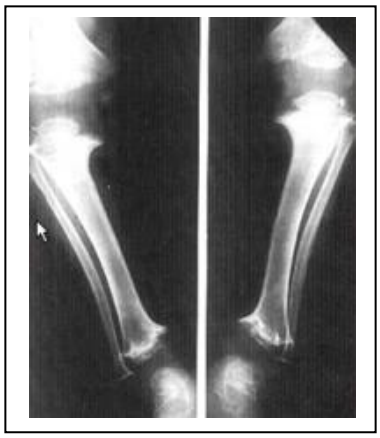
*Normal Height or Slight Short

*NO SKULL AFFECTION

- Over Growth of fibula *"much longer than tibia distally"*

"Remember Hypo.. Fibula more Hypo"

PseudAchondroplasia



…….. ***As Achondroplasia*** +

*NO SKULL AFFECTION

* **E**piphyseal & **M**etaphyseal Abnormality

D.D.: ***Metaphysis wide & irregular*** Like ***Rickets*** ,

But other manifestation + No skull affection = ***PseudoAchond***.

Differential TERMS

- **Rhizomelia = Short Proximal bones** → "as Humerus & femur "
- **Mesomelia = Short middle bones** → "radius&ulna – Tibia&fibula"
- **Acromelia = Shorts distal bones** → "Fingers"

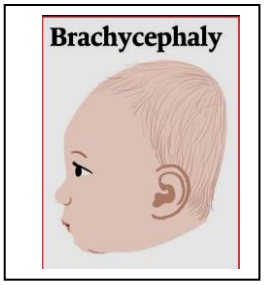
Claiedo-Carnial Dysplasia

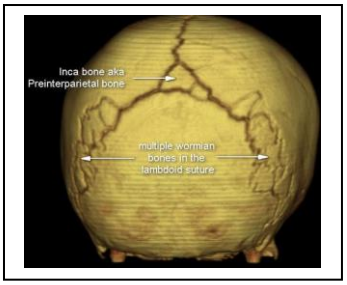
***C**liedo = **C**lavicle*
***C**ranial = Skull → manifestation mainly in skull & calvicle*

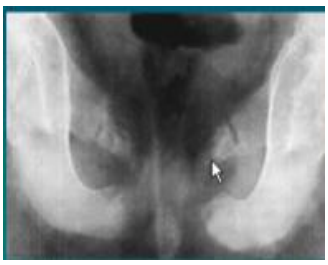
https://radiopaedia.org/articles/cleidocranial-dysostosis

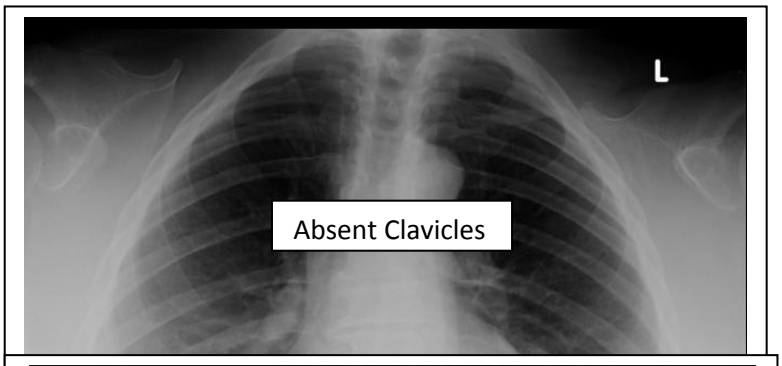
- ***Rare*** - Autosomal Dominant - 30% Gene Mutation "*CBFA1 gene*"

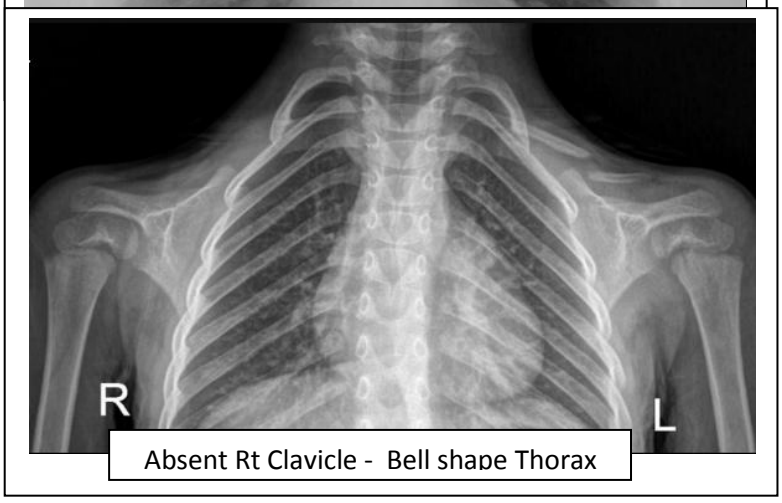
General	Ossification Abnormality *"Delay appearance of ossification center eg. Pubic"*
Skull	**-Brachecephaly:** *premature fusion of the coronal suture* → small A-P diameter **- Wormian bones :** multiple small bones in-between sutures
Clavicle	-Absent all or outer part → **High scapula**
Chest	-Narrow upper diameter → **Bell shape** Thorax -over number ribs "*Supernumerary* → Cervical or Lumber ribs"
Spine	-Biconvex vertebral
Tooth	Abnormal "count & direction"







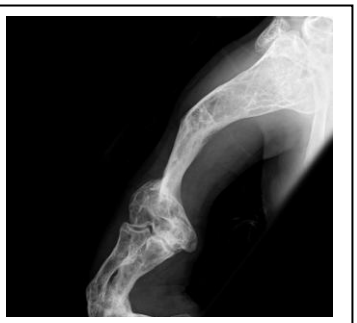


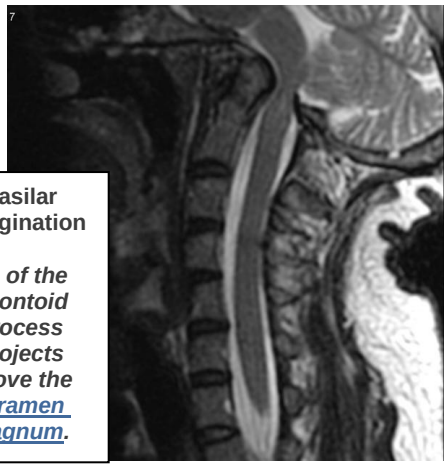


Osteogenesis Imperfecta

- Rare 1 / 15000
- Genetic disorders of **collagen type I** production
  → Osteoporosis & Fragile Bones
- **Types**: -**T**arda "Mild" -**S**ever "died early"
  & *as regarding bone shape*: -*Gracile* "*thin bones*" -*Broad*
- 5 main Manifestations:

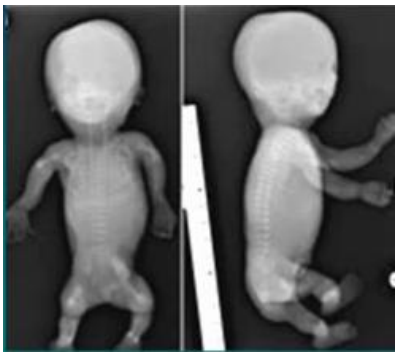
- Osteoporosis → Fractures
- Fragile bones → Thin or wide
- Lax Joints
- **D**ental Apnormality
- **D**eafness ← *Otosclerosis*
- Blue Sclera



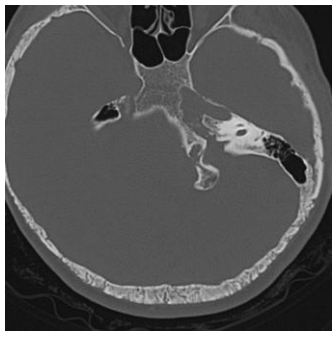


Basilar invagination

tip of the odontoid process projects above the foramen magnum.



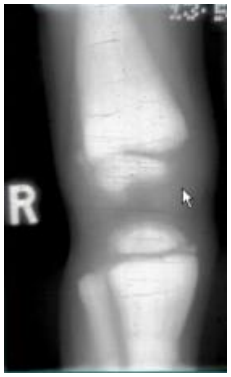
BABY GRAM

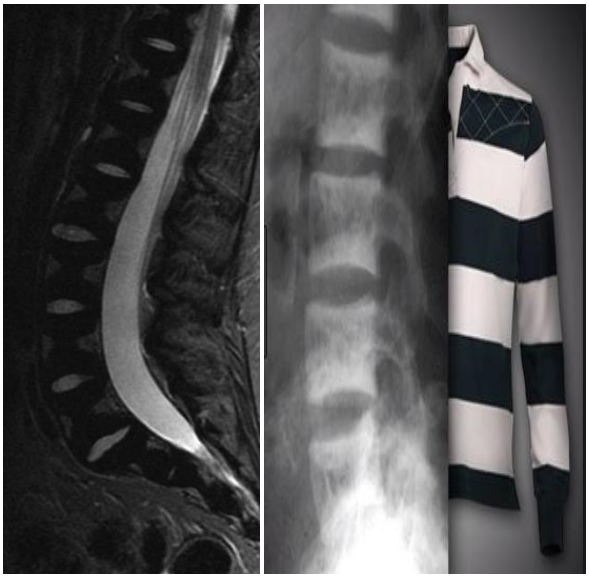


Wormian Bones in OI

OsteoPetrosis *(Marble Bone Disease)*

- Uncommon hereditary disorder ← defective osteoclasts
- →**Bones** : Sclerotic & Thick *but* Weak & Brittle
- Manifestations:
  - General increased bone density
  - **F**ractures
  - **F**lask shaped long bones
  - Bone within bone
  - **"Rugger Jersey "** Spine : *sclerotic end plates*

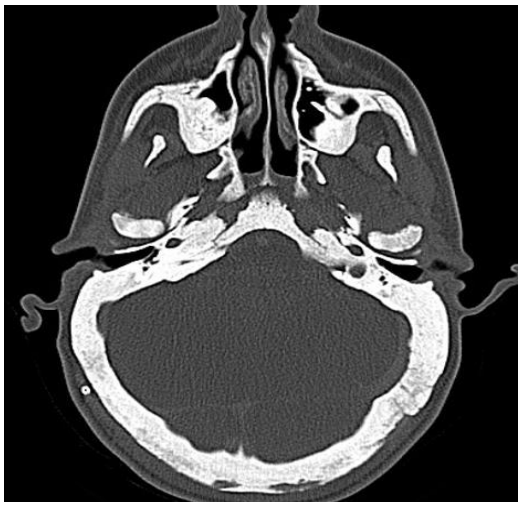




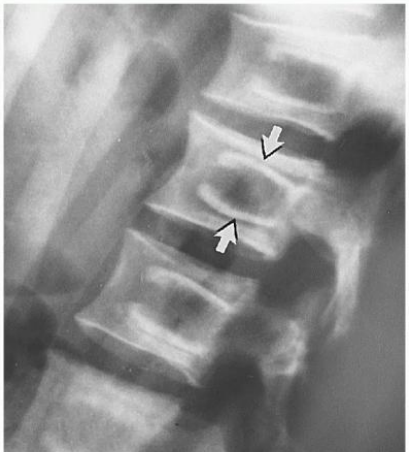
C.P. -Hepatosplenomegally -Enlarged LN -Anemia -Deafness -Optic Atrophy

D.D. of O.I.:

- heavy metal poisoning (e.g. lead)
- melorheostosis
- hypervitaminosis D
- pyknodysostosis
- fibrous dysplasia of skull or face



Bone with in Bone



FIBROUS DYSPLASIA

https://radiopaedia.org/articles/fibrous-dysplasia

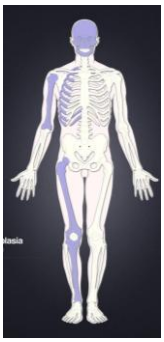
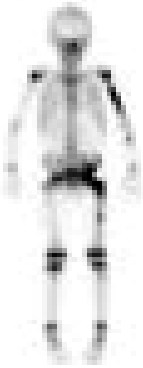
- Non-neoplastic tumour-like congenital process,
- **Presentation Age**: Young Adult (*Peak* 3 : 15 y)
- **Etiology** : Unknown …….
- **Pathology**: Medullary bone replaced by **fibrous tissue**

  → Metaphyseal cystic lesion with "*ground glass veiling*"

  + Bone deformity
- **Types**:

  - **Mono**stotic "*single Bone*" 70:80% → Ribs > Proximal Femur > Tibia > ……

  - **Poly**stotic *"Multiple Bones"* 20:30% → Femur 91% > Tibia 81% > Pelvis > …….. least spine.

Monostotic	***Polystotic***
	

⇨ **C.P.:**

-Incidental +/- Endocrine Abnormality

- Painless bone remodeling

-Adjacent structures compression

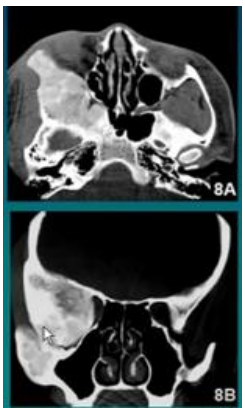
N.B. ***Monostotic Never progress to Polystotic***

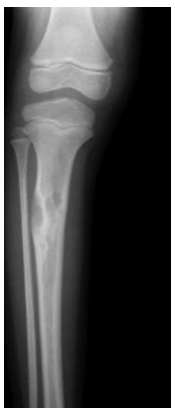
⇨ **Treatment :**

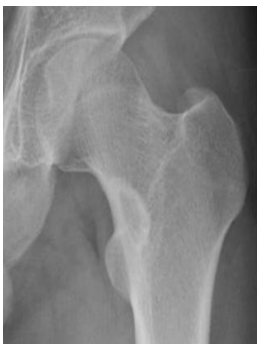
- Usually No ttt
- Surgical decompression ← Mass effect

D.D. -Pagets disease - NF 1 - NOF - Adamantinoma - Enchondromas

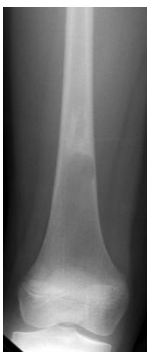
X ray **CT**	• **Ground-glass** matrix * May be completely lucent (cystic) or sclerotic • well circumscribed lesions * **No periosteal reaction** • rind sign
MRI	Marked variability in the appearance
Bone scan	increased tracer uptake on Tc^{99} bone scans



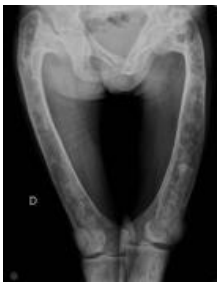




Ring Sign

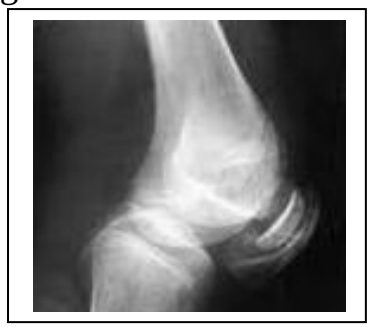


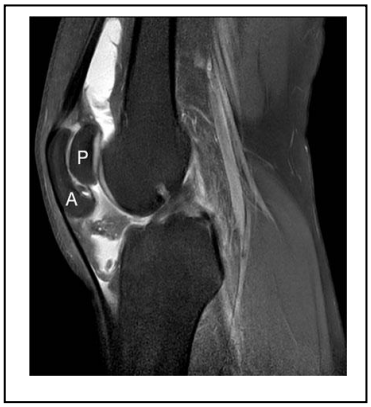
Polystotic F.D.



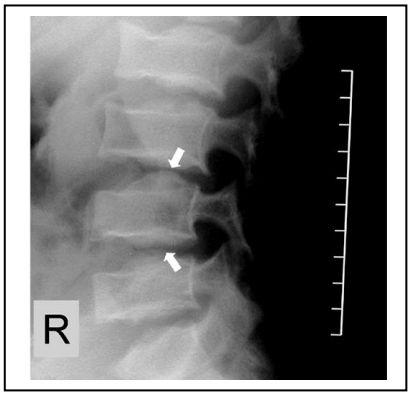
Multiple Epiphyseal DYSPLASIA

- Autosomal Dominant - Dwarfism
- Diffuse **epiphyseal** changes in long bones :
  - Delayed
  - Smaller
  - Irregular
  - Fragmented
- This lead to → Premature O.A.
- **Double Layered Patella**





Spondylo Epiphyseal DYSPLASIA

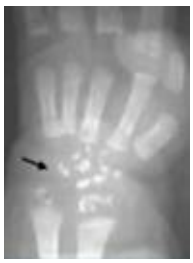


As Multiple Epiphyseal Dyspkasia

\+ **Platysponyly** = ***Redced Hight + irregular endplate***

Punctate Epiphyseal DYSPLASIA

- **Stippled Epiphysis** →replaced by multiple Ca dots
- **Rhizomilia** → Short Proximal bones
- Cartilage calcification eg. *Trachea , Bronchi*
- **Coronal Spine Cleft** ← PATHOGNOMONIC

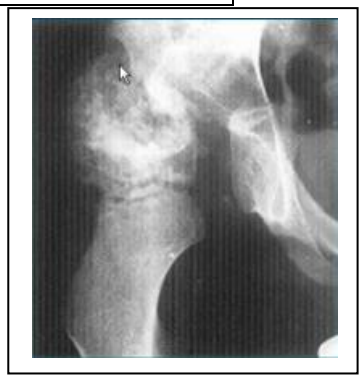


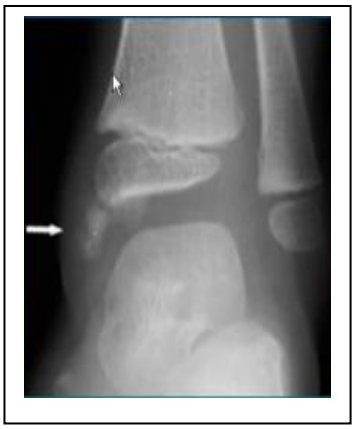
Stippled Epiphysis

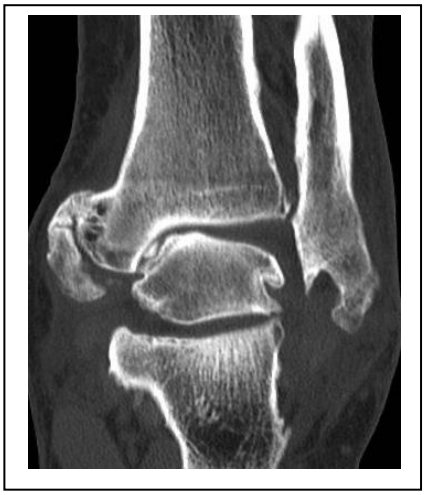
HEMIMELICA Epiphyseal DYSPLASIA

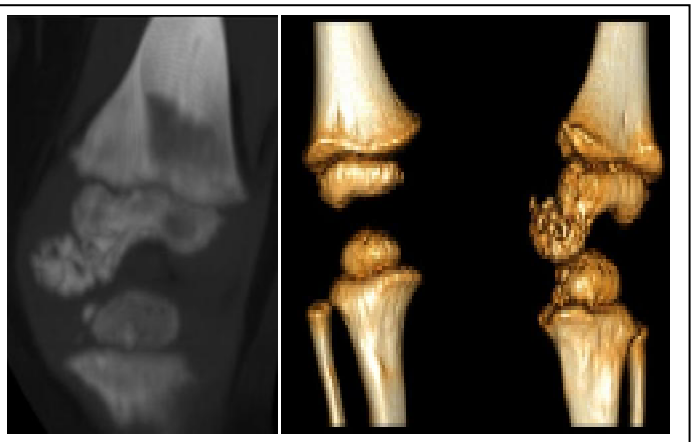
Or Trevor's Disease

- Irregular **over growth** of epiphysis or apophysis At one limb **on One side**.
- Occurs in L.L. > U.L.
- **D.D.** : **Osteochondroma** ← *arise from metaphysis*



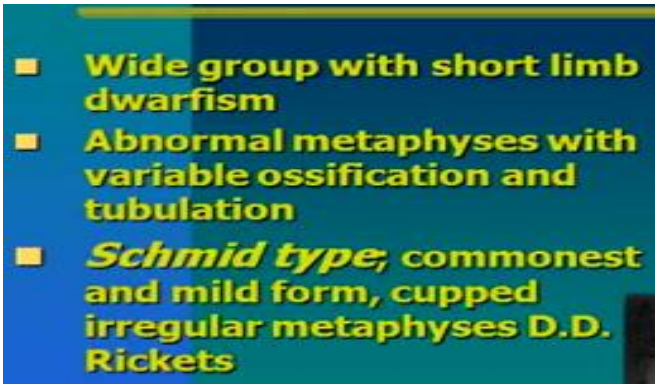


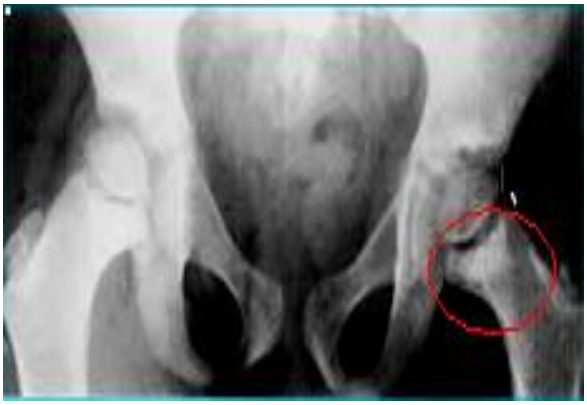


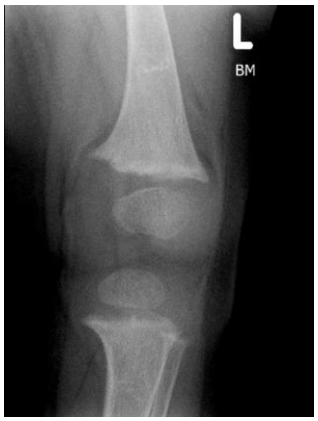


"Special Character"		
Epiphyseal Dysplasia	**Multiple**	**Double Layered Patella**
	Spondylo	Multiple + **Platysponyly**
	Punctate	**Stippled Epiphysis** *** Coronal Spine Cleft**
	Hemimelica	Epiphyseal overgrowth of one limb at one side

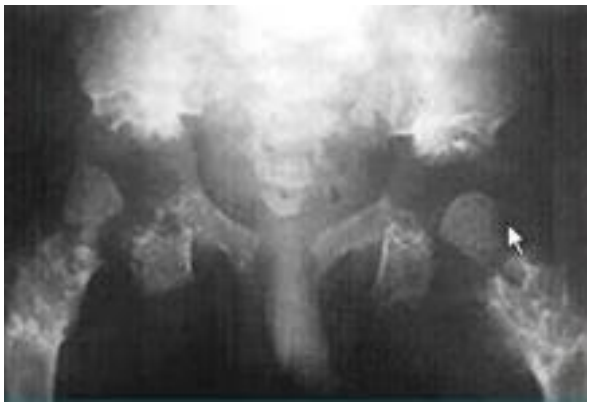
METAPHYSEAL CHONDRODYSPLASIA







RICKETS

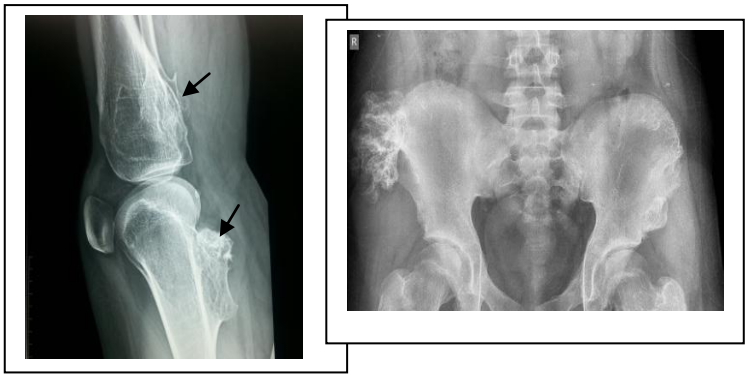


Sever Form of Metaphyseal Chondroplasia

DIAPHYSEAL ACLASIS

"Hereditary Multiple Exostosis"

https://radiopaedia.org/articles/hereditary-multiple-exostoses

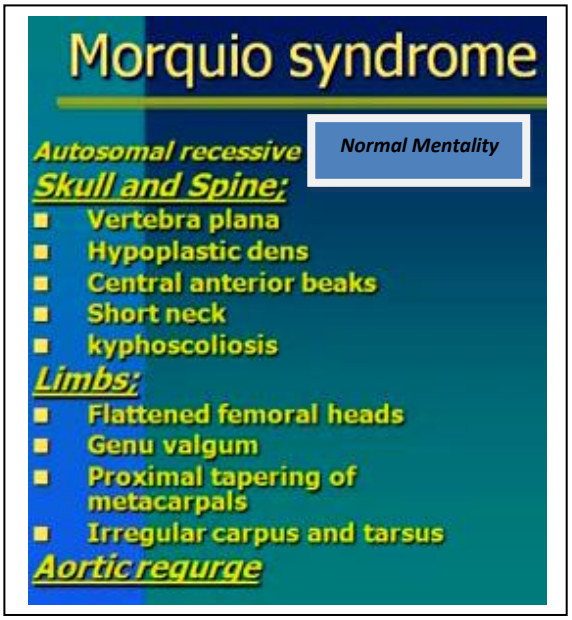


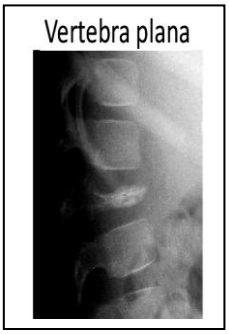
- Familial or 40 % Mutation
- Multiple Osteochondromas
- Arise in Metaphysis of,
  progress to diaphysis on growth
  So …..←"*Diaphyseal Misnomer*"
- **Long bone**
- Can involve **Pelvis** , **Vertebra** & **Scapula**
- Sessile or pedunclated , Directed away from joint
- **10 %** malignant transform→***Chondrosarcoma***

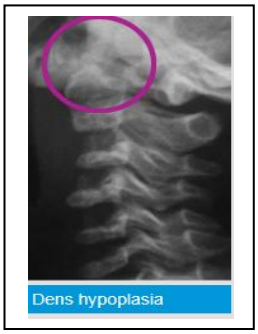
MUCOPOLYSACCHARIDOSIS

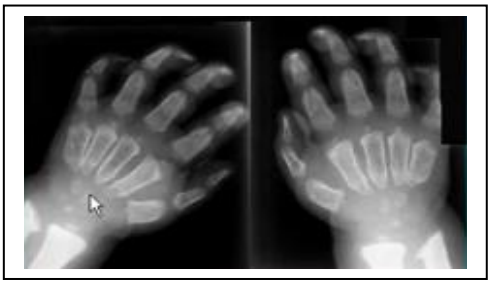
- Abnormal deposition of ***Mucopolysccharides*** in skeleton & soft tissue.
- **C.P.:** -Mental Retardation - Corneal Clouding - Skeletal Changes -Organomegally
- **Subtypes :** many subtypes , main 3 types

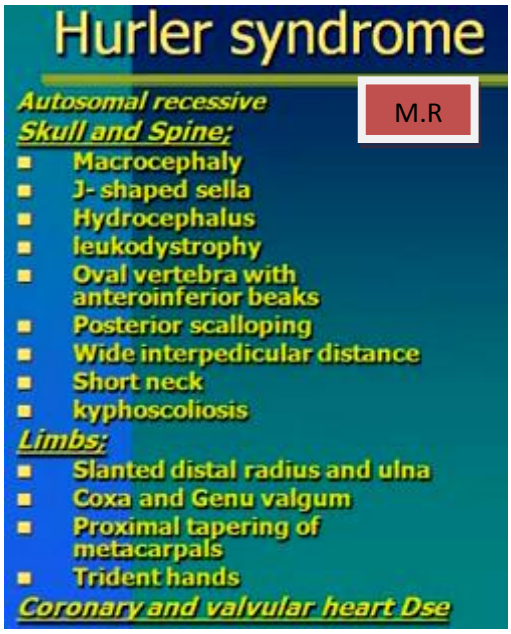
1- Morquio syndrome "*Mild form***" 2-Hurler Syndrome 3-Hunter Syndrome**

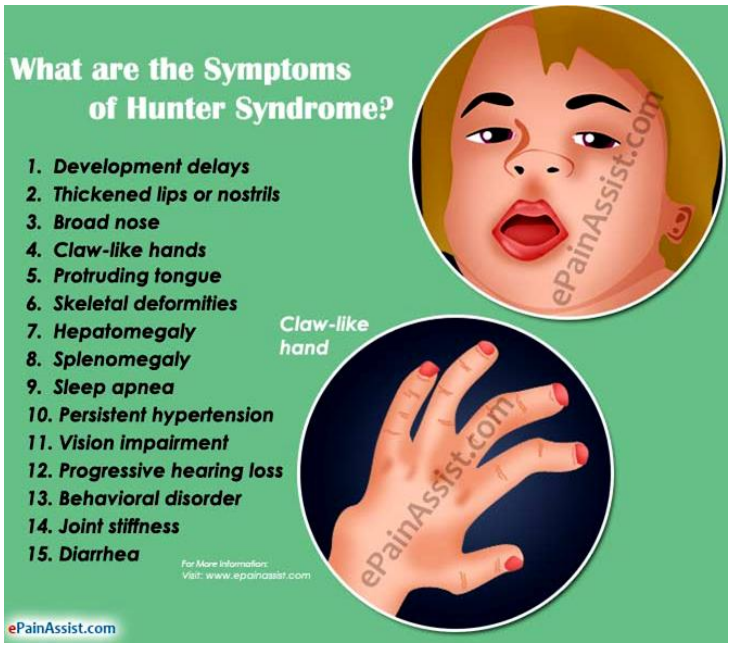




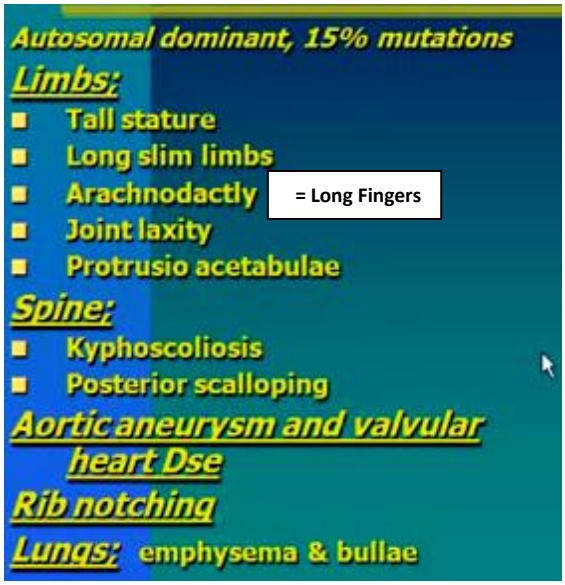


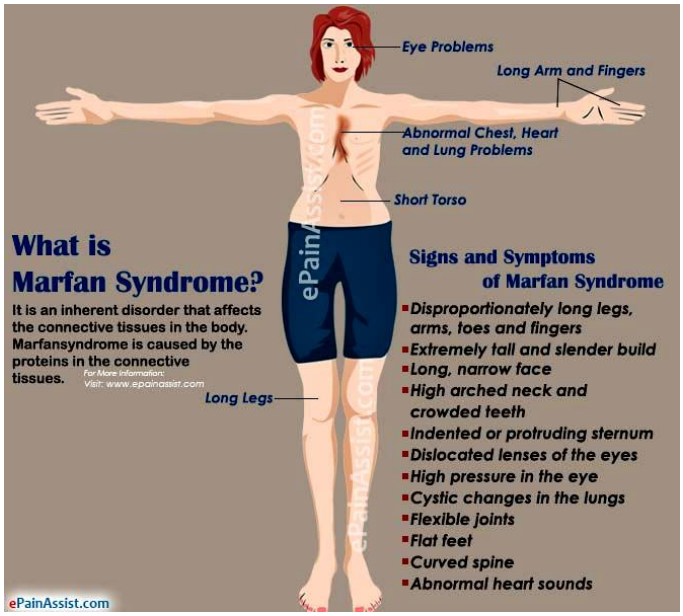



Hurler syndrome
M.R
Autosomal recessive
Skull and Spine:
Macrocephaly
J- shaped sella
Hydrocephalus
leukodystrophy
Oval vertebra with anteroinferior beaks
Posterior scalloping
Wide interpedicular distance
Short neck
kyphoscoliosis
Limbs:
Slanted distal radius and ulna
Coxa and Genu valgum
Proximal tapering of metacarpals
Trident hands
Coronary and valvular heart Dse

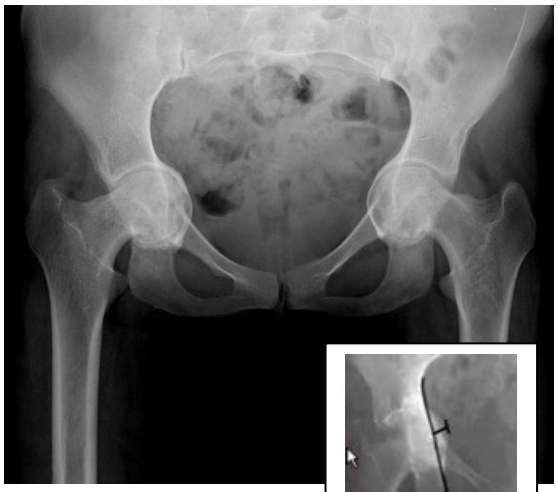

What are the Symptoms of Hunter Syndrome?
1. Development delays
2. Thickened lips or nostrils
3. Broad nose
4. Claw-like hands
5. Protruding tongue
6. Skeletal deformities
7. Hepatomegaly
8. Splenomegaly
9. Sleep apnea
10. Persistent hypertension
11. Vision impairment
12. Progressive hearing loss
13. Behavioral disorder
14. Joint stiffness
15. Diarrhea
Claw-like hand
ePainAssist.com

MARFAN SYNDROME



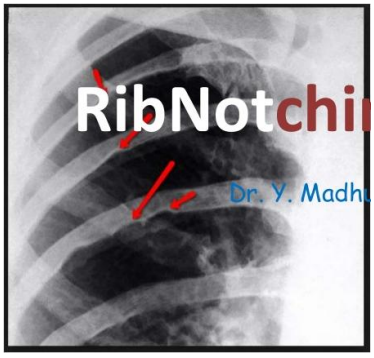


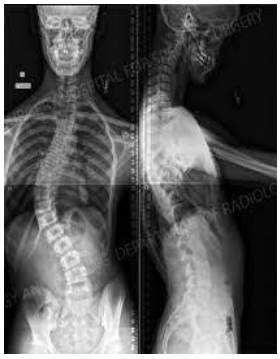
https://radiopaedia.org/articles/marfan-syndrome

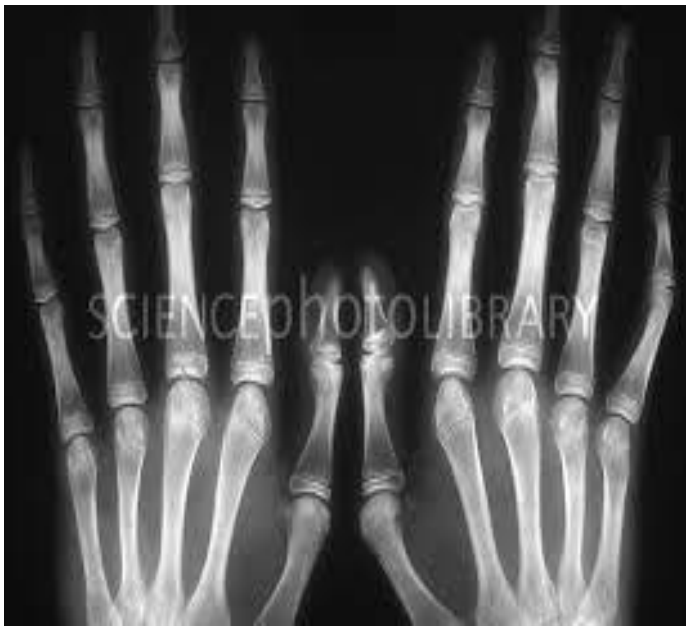


Protrusio Acetabulae :

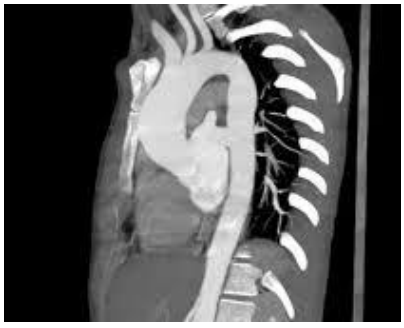
Femur head bulging inward





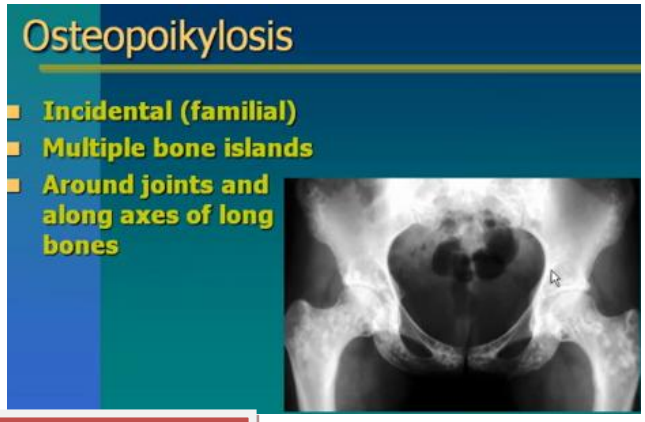


Wide Aortic Base



NB	DYSPLASIA	DYSTOSTOSIS
	Diffuse abnormal growth of bones – either cartilaginous or osseous components	Specific bones abnormal ossification of individually or in combination
	Modeling usually affecting all bones in a similar fashion	Remodeling usually affecting some bones & may be unilateral or peripheral
	e.g:- ❑Osteogenesis imperfecta. ❑ Multiple epiphyseal dysplasia. ❑Achondroplasia	e.g:- ❑cleidocranial dysostosis. ❑ craniofacial dysostosis. ❑Dysostosis multiplex

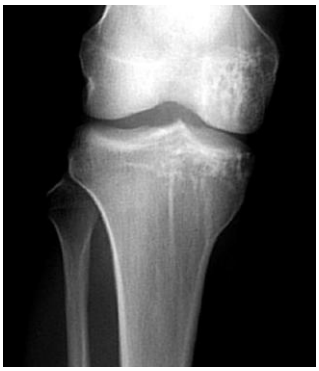
OTHERs Mimicking Dysplasias



Exclude Sclerotic Mets By Bone scan

Osteopathia Striata

- Rare
- Asymptomatic
- Longitudinal striation
- Long bones esp Lower Limb



SOURCES

- *Lecture of Prof. Ahmed Wafee* https://www.youtube.com/watch?v=MlSX98yuqNg&t=309s
- *Lecture of Prof. Hassan Elkiki* https://www.youtube.com/watch?v=x-tRdBcVKaw
- www.Radiopaedia.org

By A.M.Abodahab
Oct 2017