My Bumpy bones

They're bumpy

They're lumpy

At times I feel frumpy

With bones that behave

As if I'm not not in Charge…

SKELETAL DYSPLASIA

Skeletal: Bone

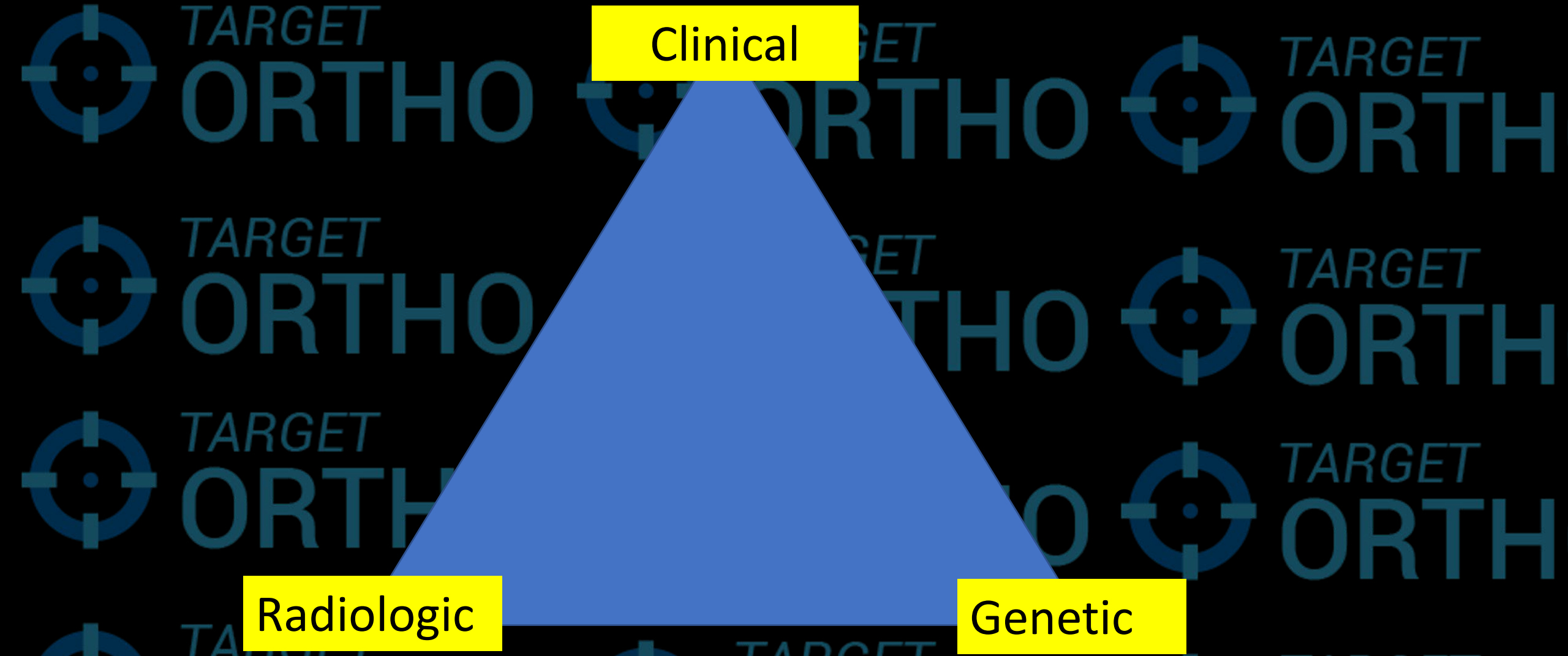
Dys: Disordered

Plasia: Formation

SKELETAL DYSPLASIA

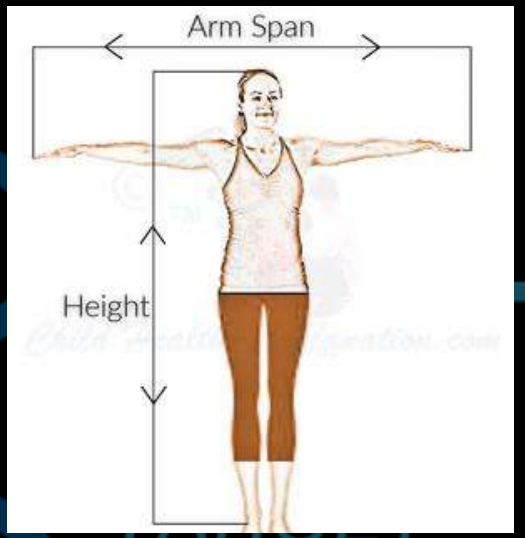
Disorder of cartilage and bone growth resulting in abnormal shape and size of skeleton and disproportion of long bones, spine and skull

=DISPROPORTIONATE SHORT STATURE



DISPROPORTIONATE SHORT STATURE

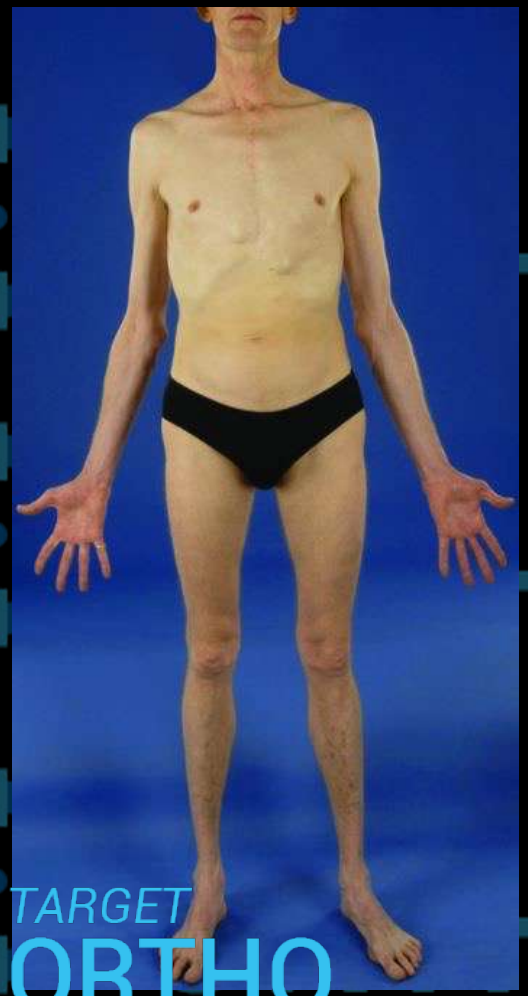
TRUNK- EXTREMITY RATIO:

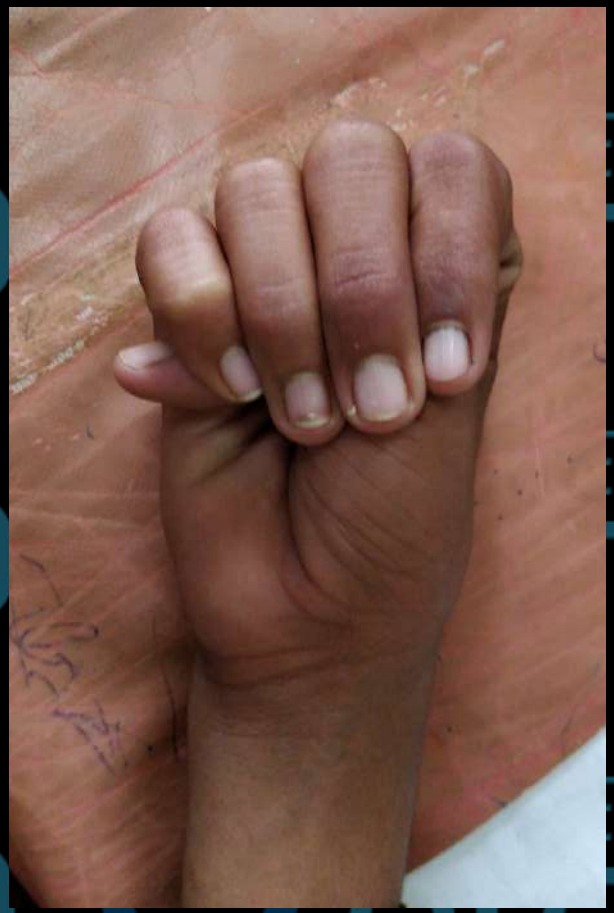


1.NORMAL TRUNK AND SHORT EXTREMITIES (ACHONDROPLASIA)

2.NORMAL LIMBS AND SHORT TRUNK (S.E.D TARDA)(MPS)

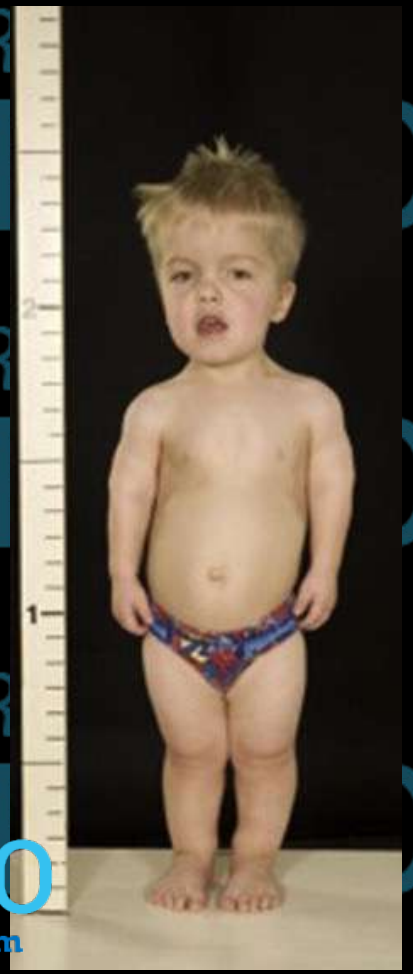
3.LONG TRUNK AND LONG LIMBS (MARFANS)





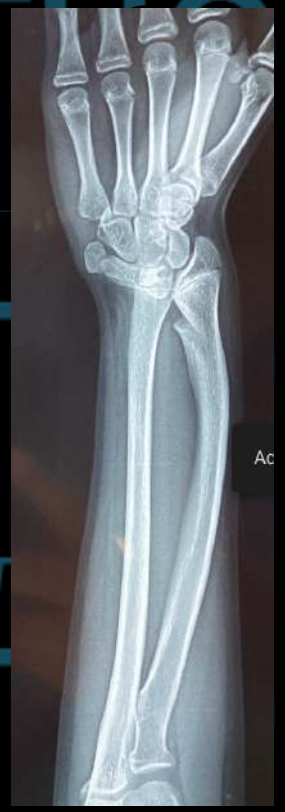
LIMB- SEGMENT RATIO

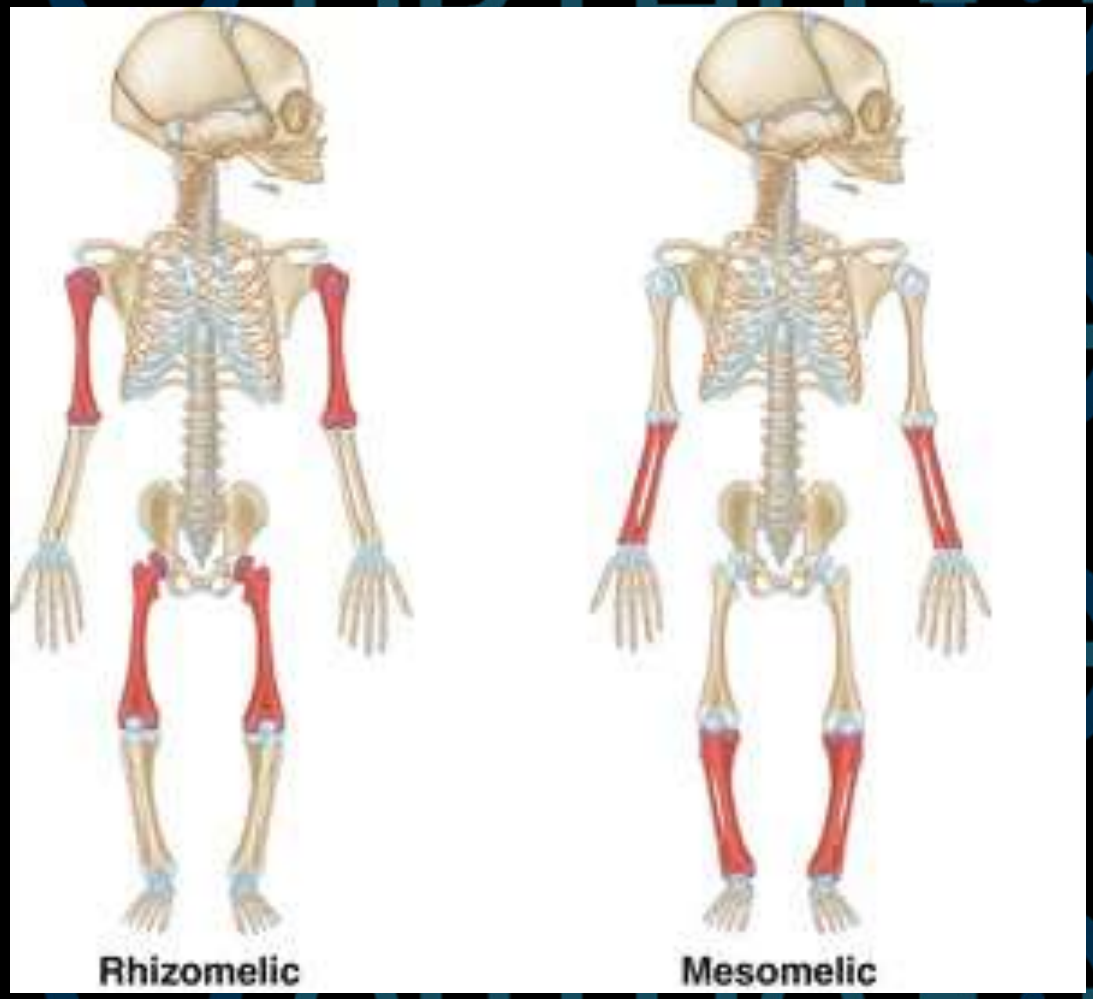
RHIZOMELIC	ACHONDRODYSPLASIA SED



LIMB- SEGMENT RATIO

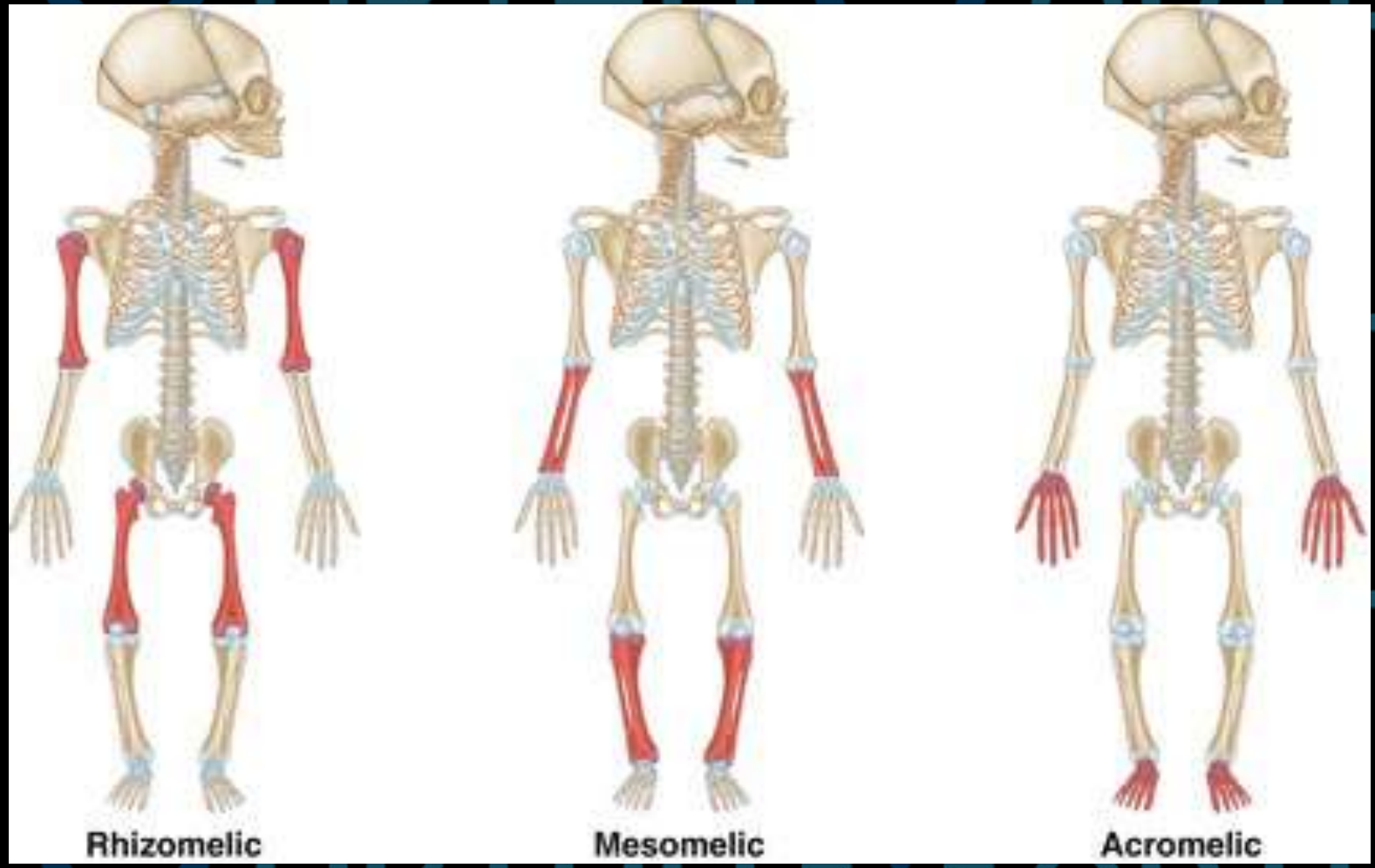
MESOMELIC	LERI WEIL DYSPLASIA

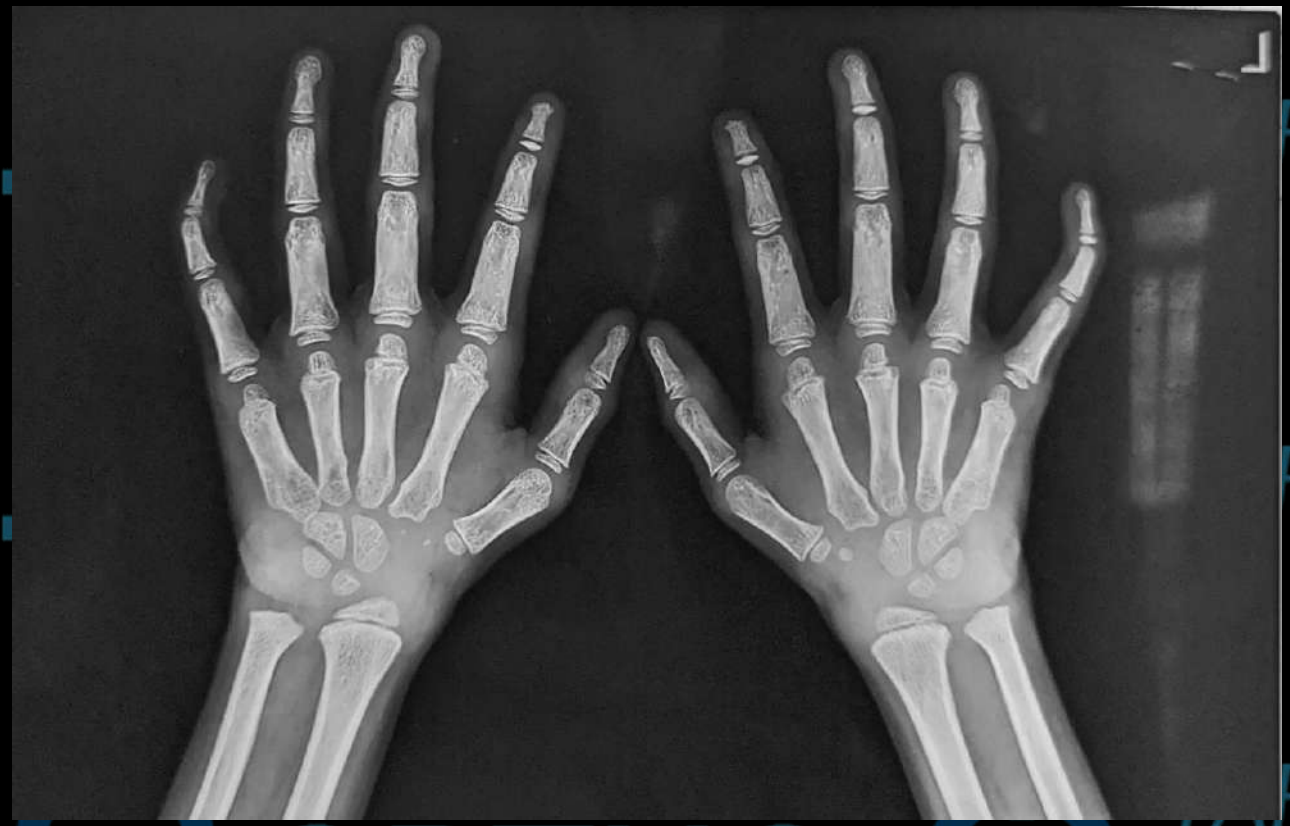


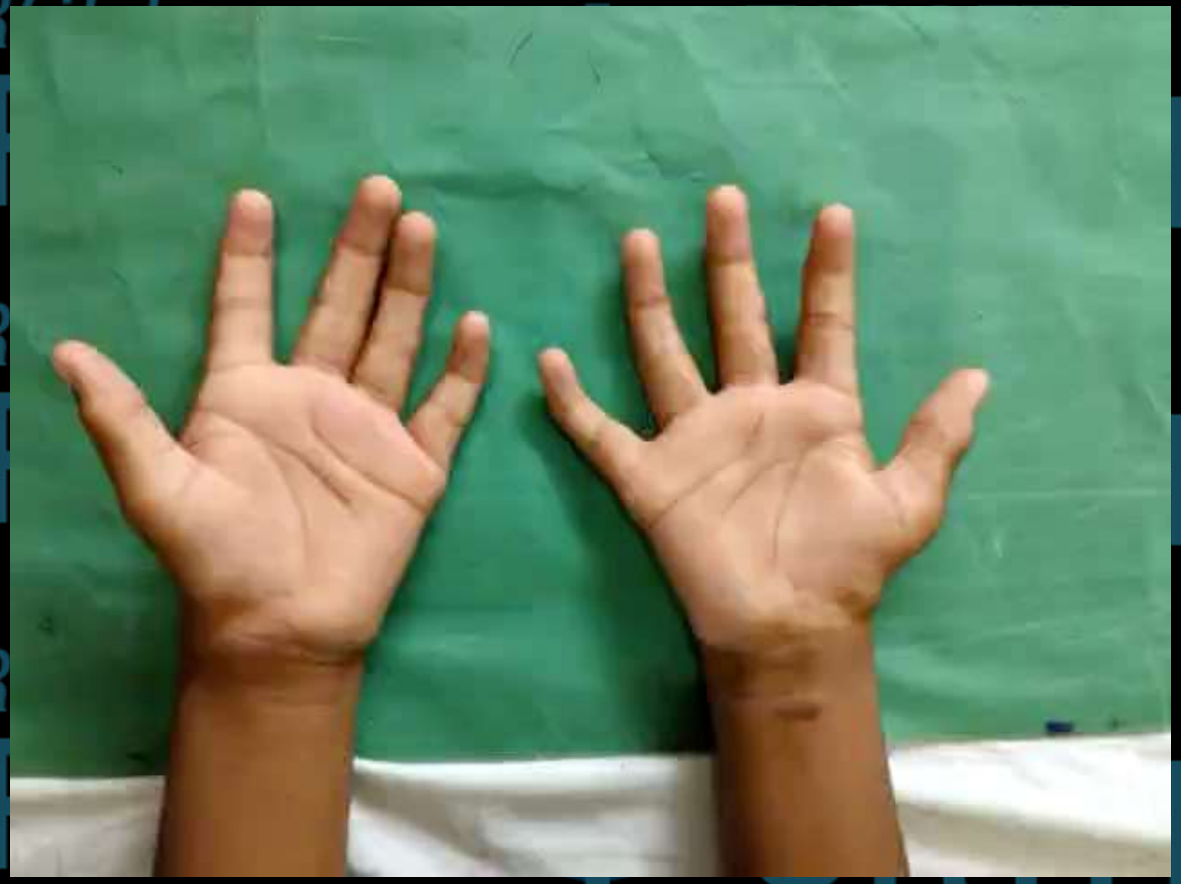


LIMB- SEGMENT RATIO

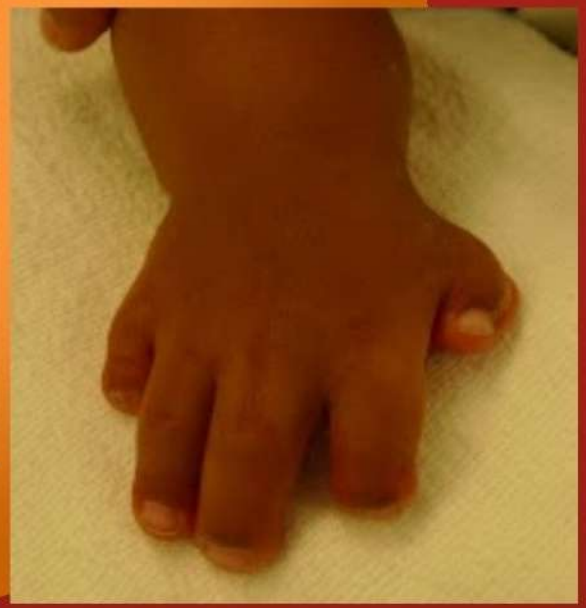
RHIZOMELIC	ACHONDRODYSPLASIA SED
MESOMELIC	LERI WEIL DYSPLASIA
ACROMELIC	ACRODYSOSTOSIS





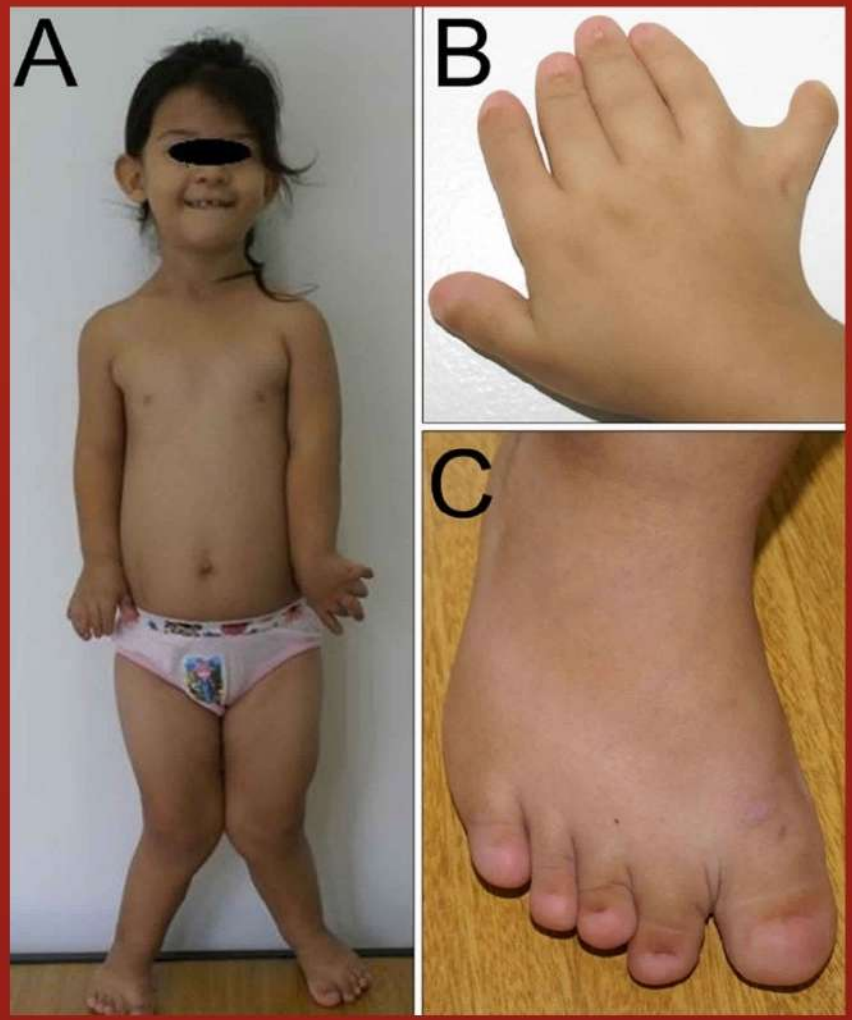


ACROMESOMELIA



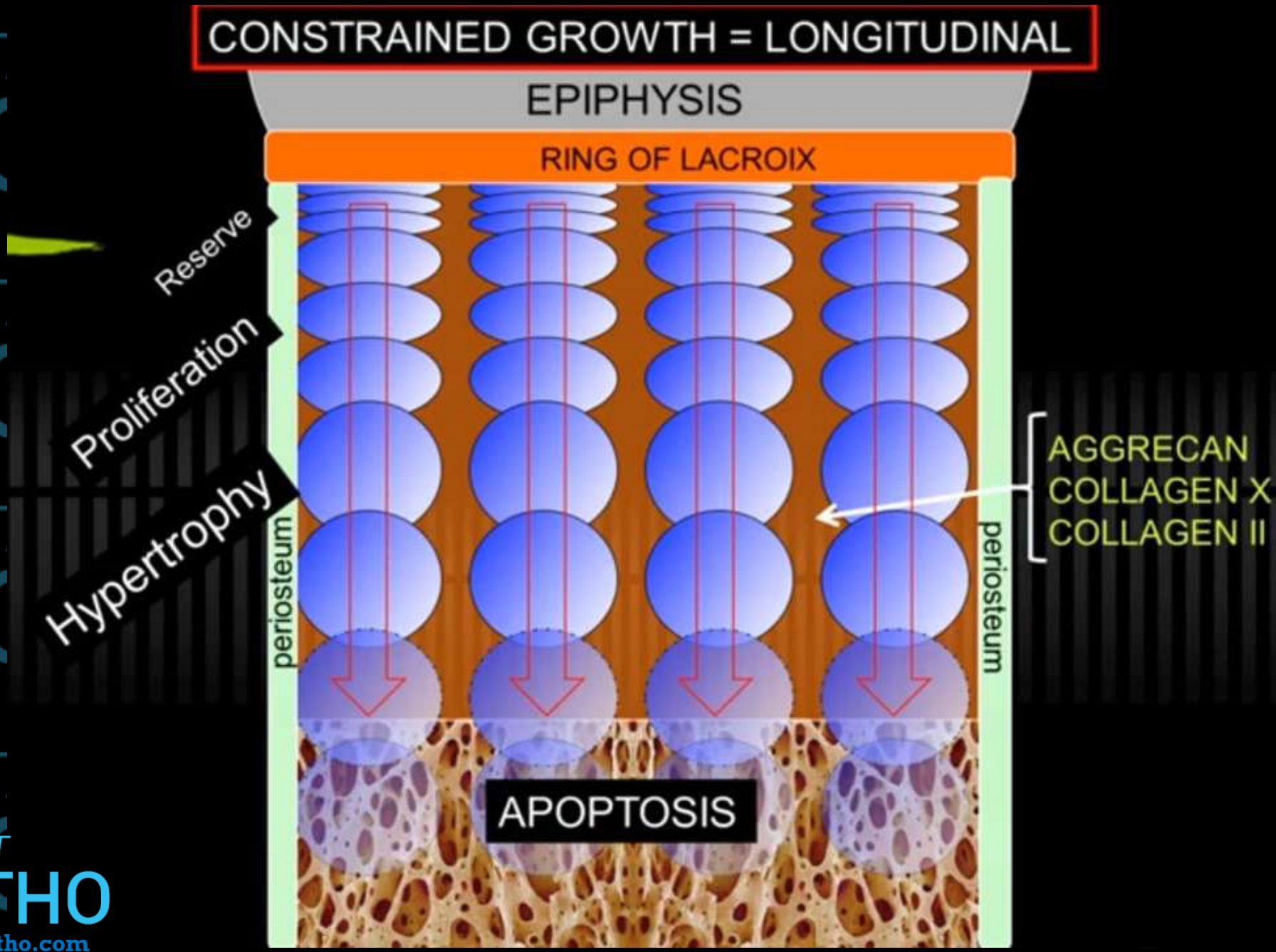
ROBINOW SYNDROME

Pic courtesy http://congenitalhand.wustl.edu/2012/05/robinow-syndrome.html



ELLIS VAN CREVELD

https://healthjade.net/ellis-van-creveld-syndrome/

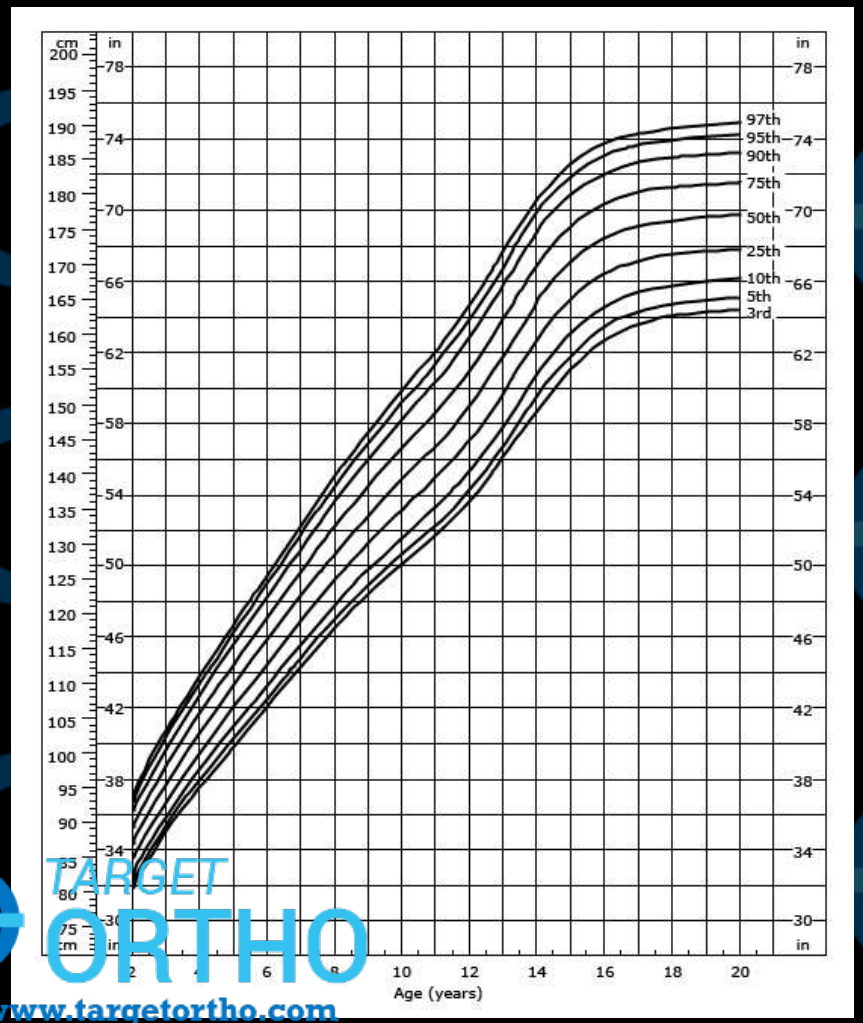


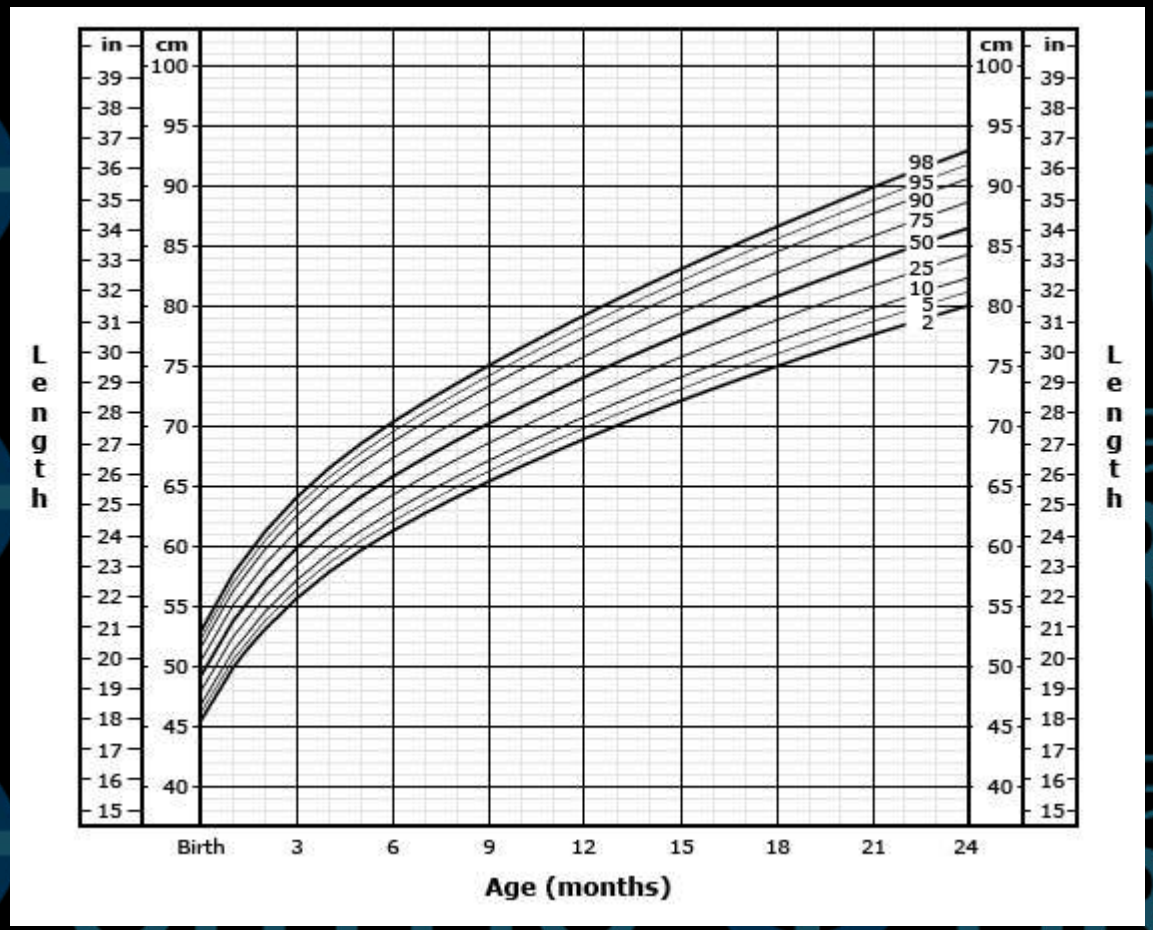
SKELETAL DYSPLASIA

CLINICAL FEATURES

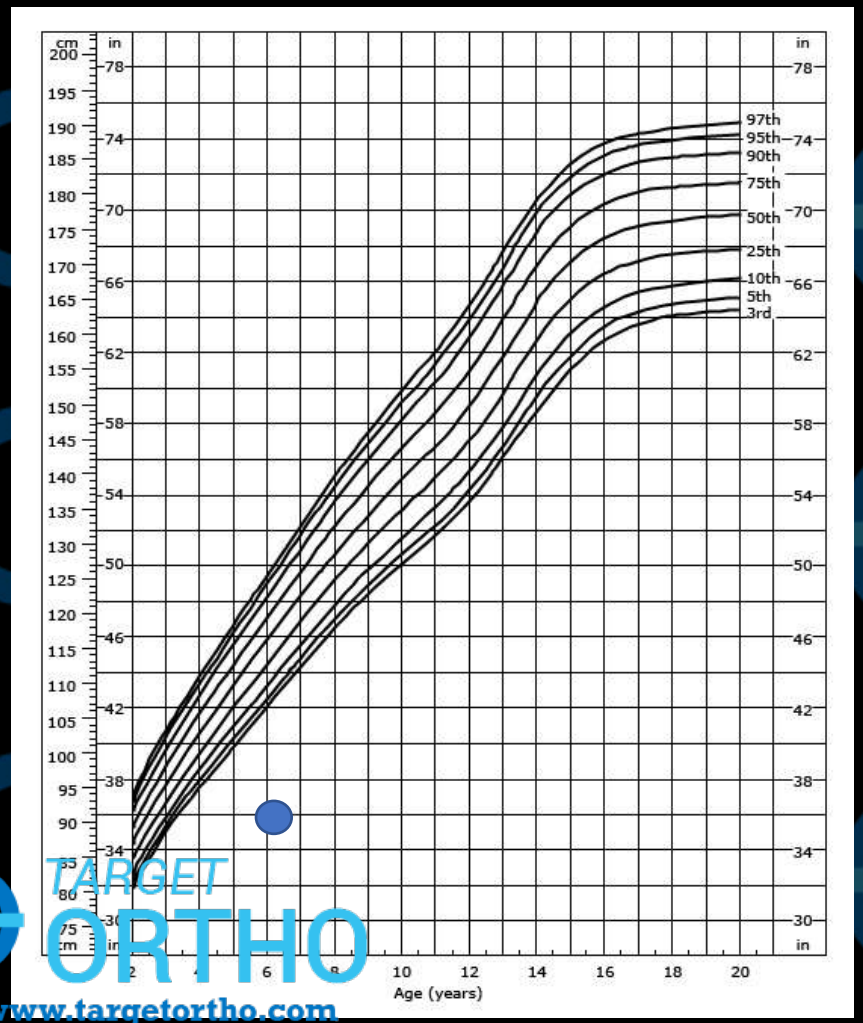
1. SHORT STATURE-
2. DISPROPORTIONATE
3. DYSMORPHISM
4. ASSOCIATED CONGENITAL MALFORMATIONS
5. DEFORMITIES

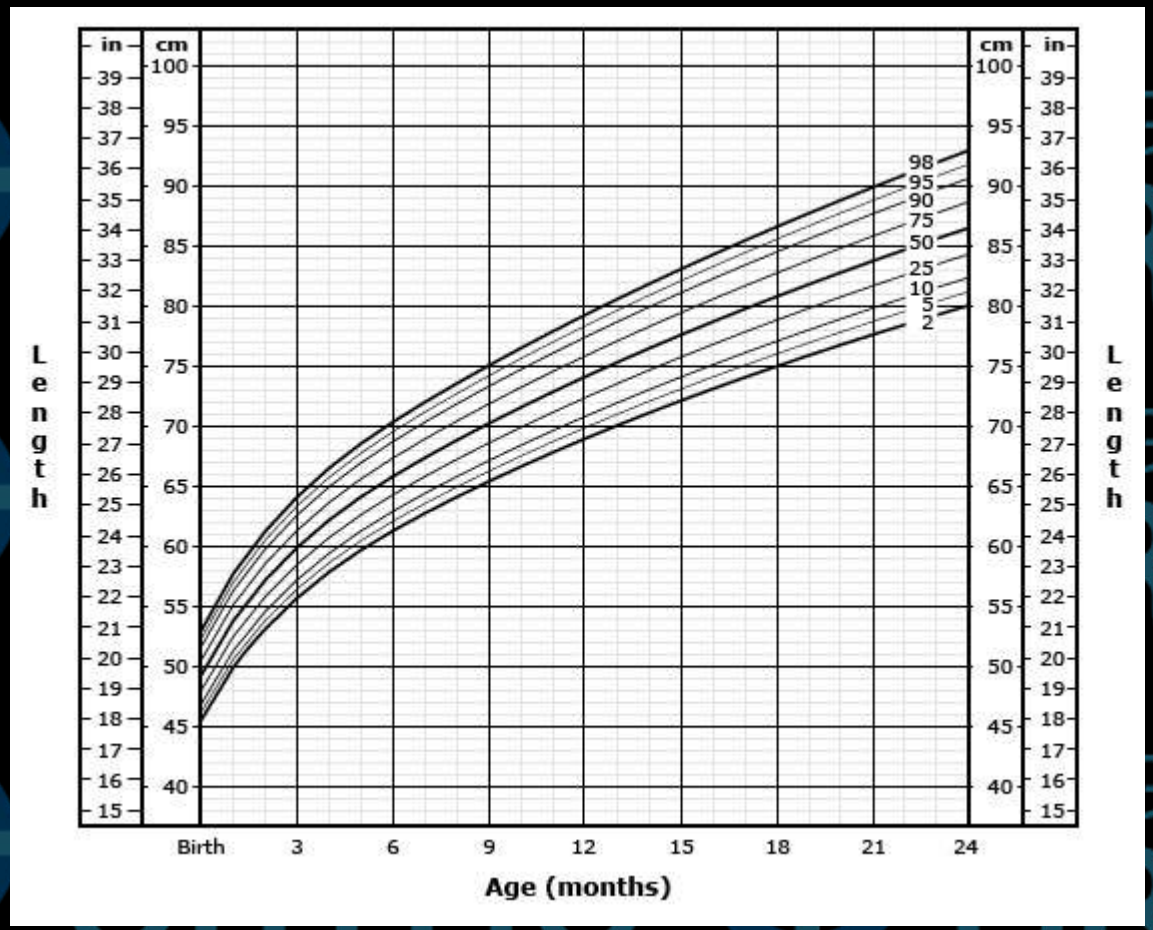
1. Short Stature





1. Short Stature

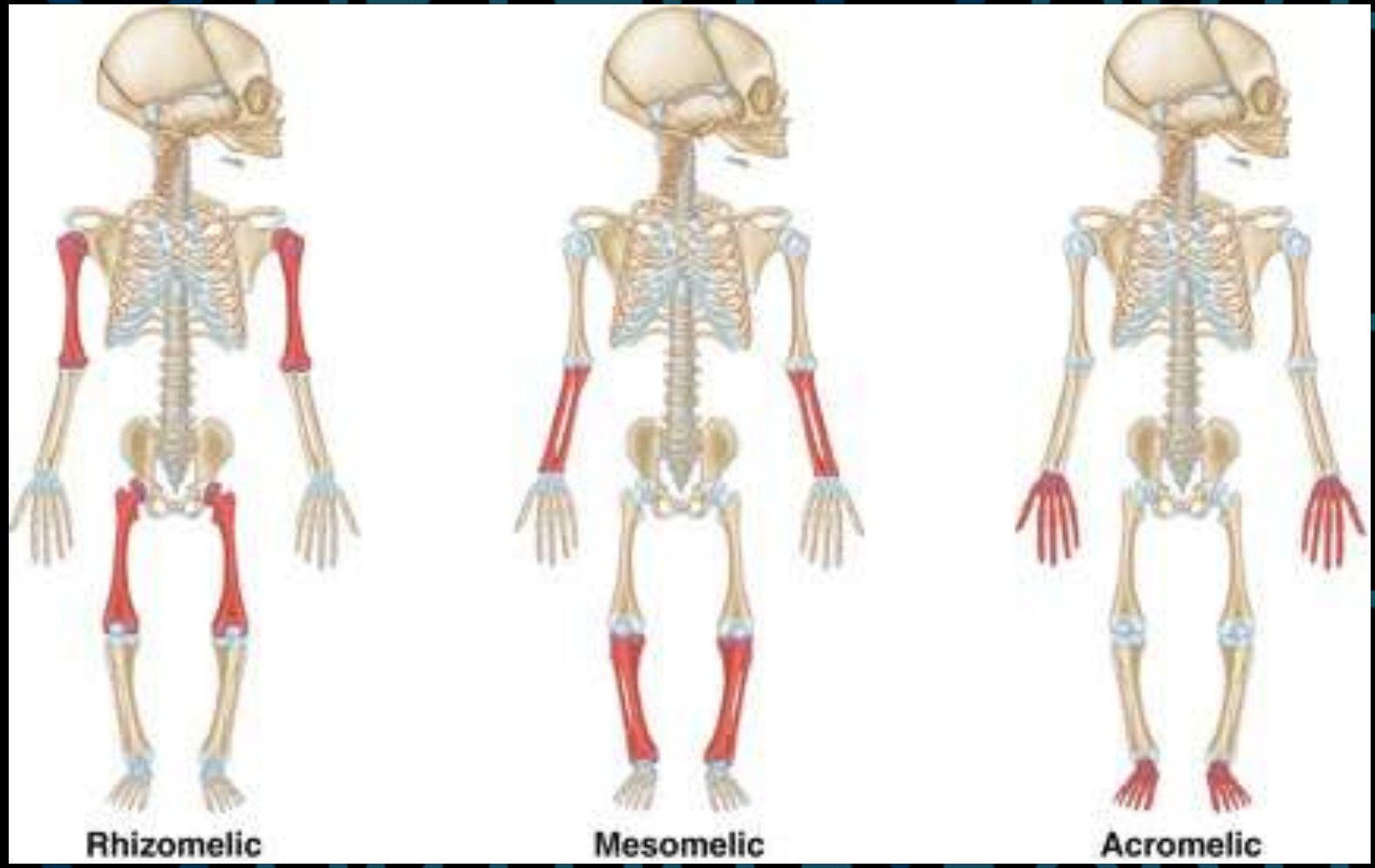


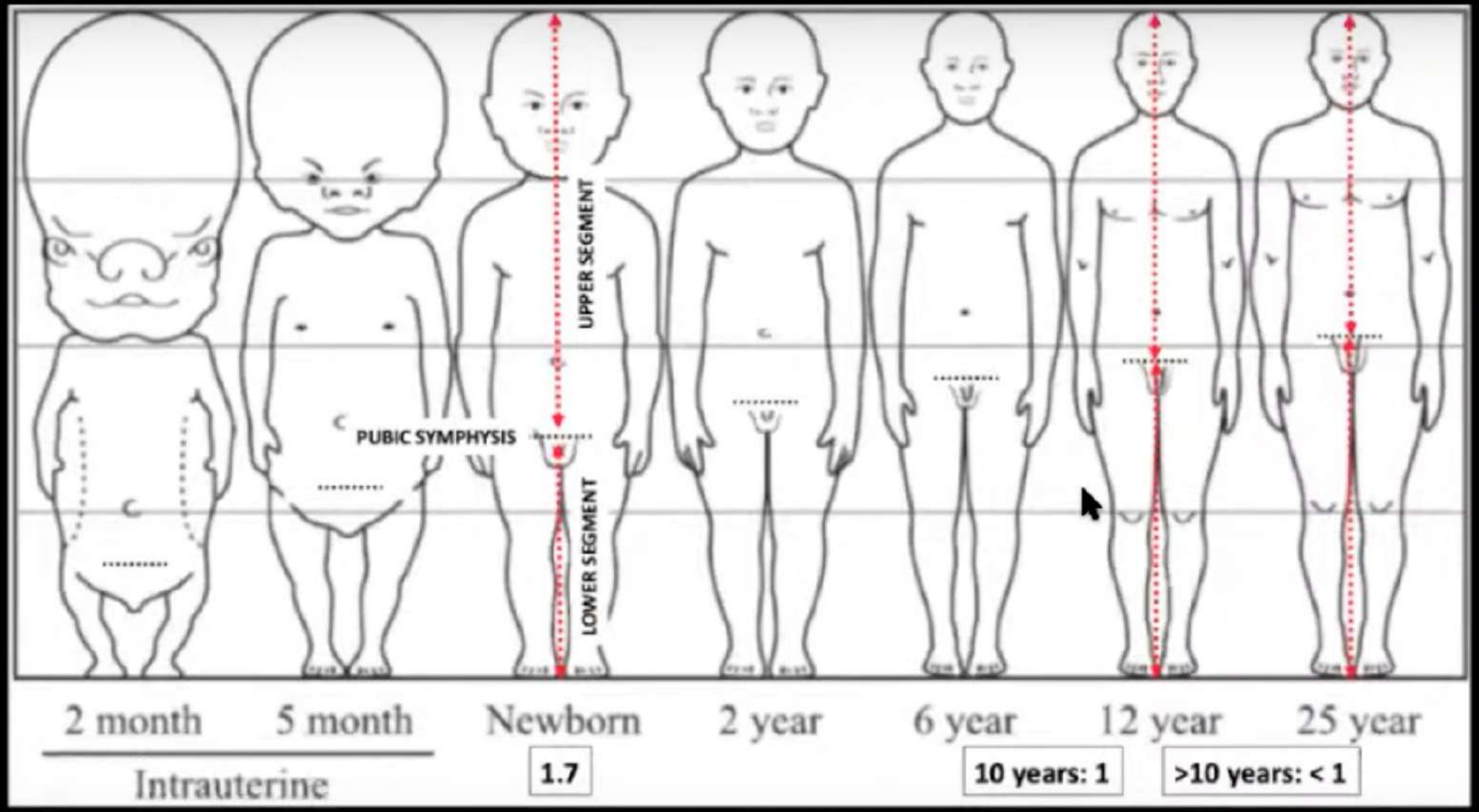


2. DISPROPORTIONATE SHORT STATURE

LIMB- SEGMENT RATIO

RHIZOMELIC	ACHONDRODYSPLASIA SED
MESOMELIC	LERI WEIL DYSPLASIA
ACROMELIC	ACRODYSOSTOSIS
MESOMELIC	ACHONDROGENESIS



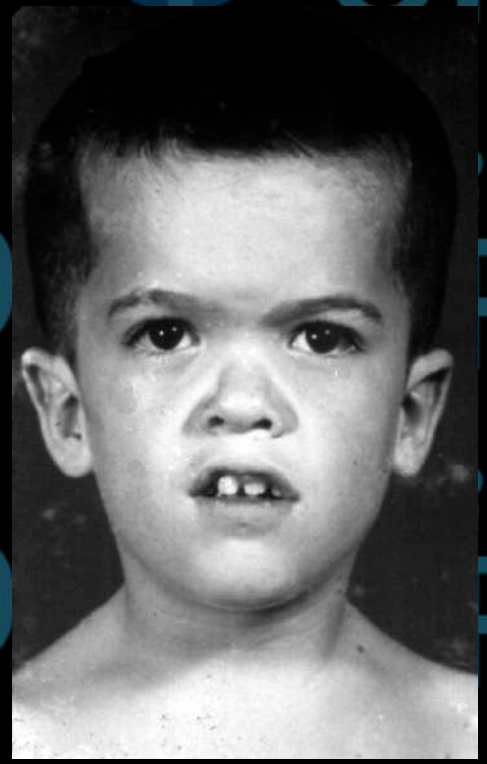


Handbook of normal physical measurements Hall and Allanson

3. DYSMORPHISM

Morphologic Variation Of Bone And Soft Tissue.

Eg..



Multiple Exostosis.

Short Broad Thumbs (M.E.D).

Depressed Bridge Of Nose (Achondroplasia)

4. DEFORMITIES

SPINAL DEFORMITIES.

LIMITED JOINT MOVEMENTS.

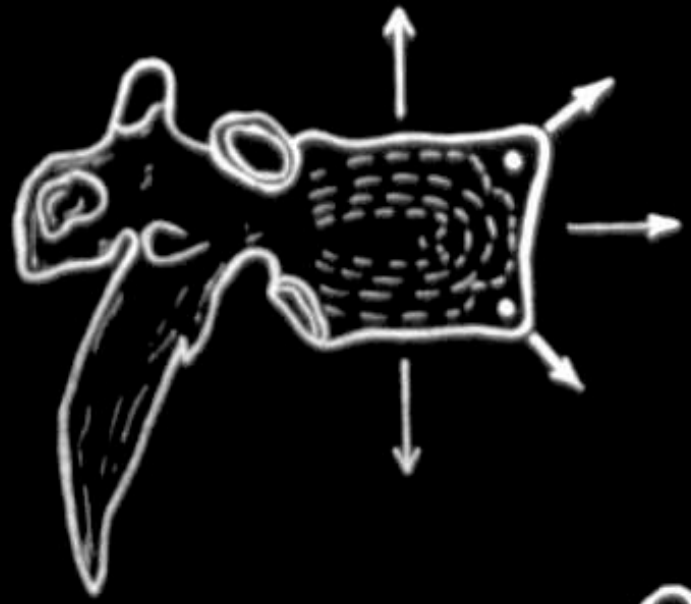
COXA VARA, GENU VARUS AND VALGUS DEFORMITIES

SKELETAL DYSPLASIAS - DIAGNOSIS

SKELETAL SURVEY X-RAYS:

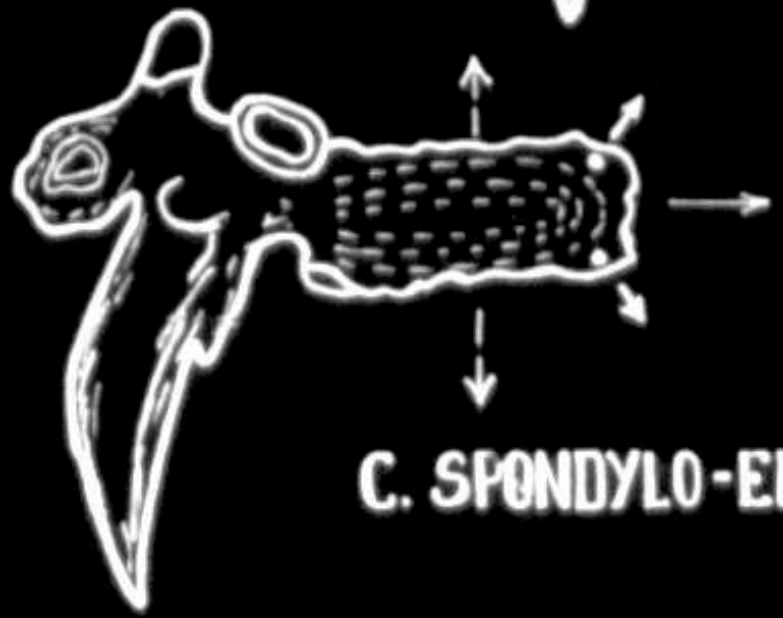
INVOLVEMENT	DISEASE CATEGORY
A + D	NORMAL
B + D	EPIPHYSEAL DYSPLASIA
C + D	METAPHYSEAL DYSPLASIA
B + E	SPONDYLOEPIPHYSEAL DYSPLASIA
C + E	SPONDYLOMETAPHYSEAL DYSPLASIA

- LATERAL SKULL
- AP PELVIS
- LATERAL LUMBAR SPINE
- AP KNEE
- AP WRIST AND HAND

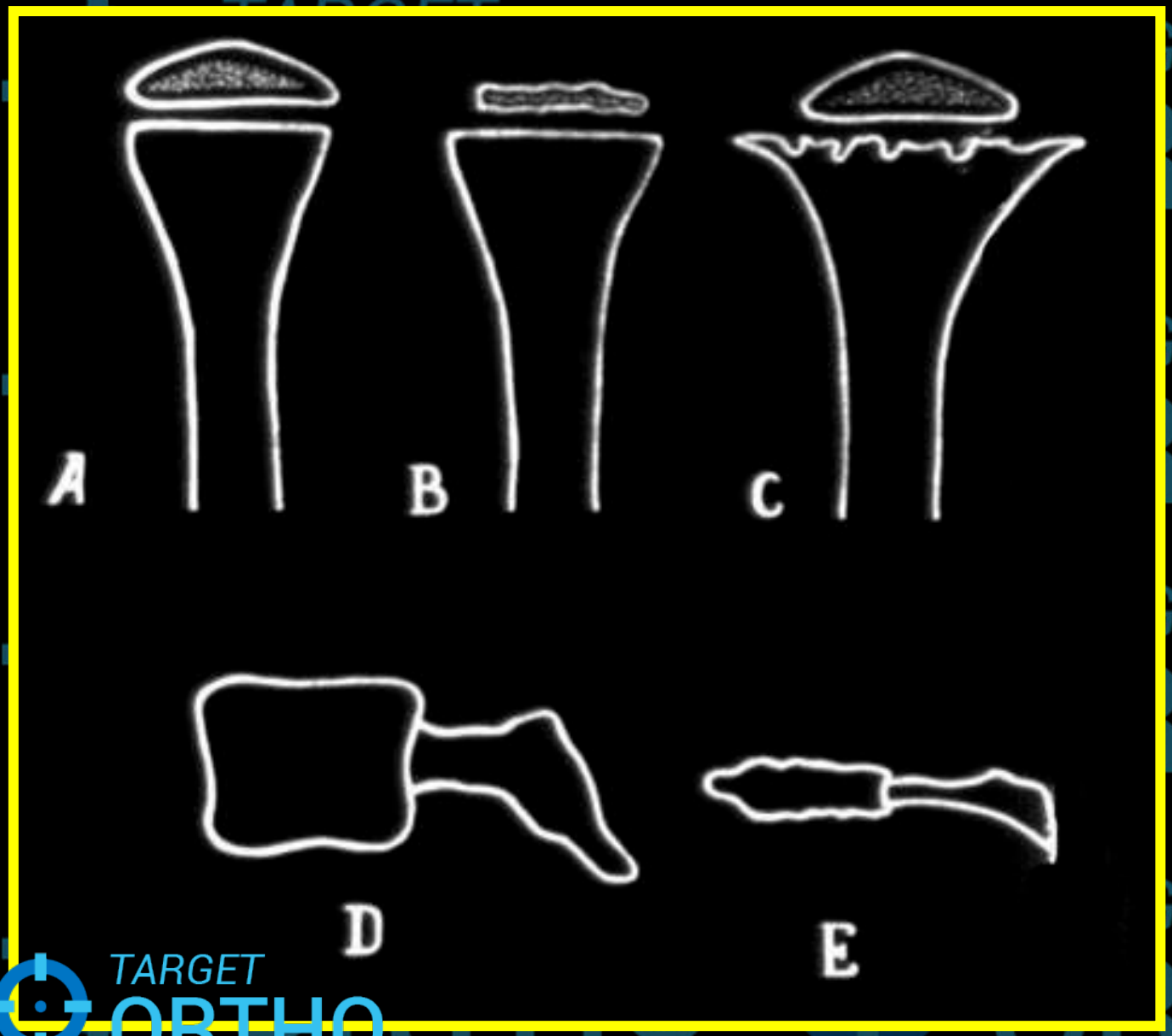


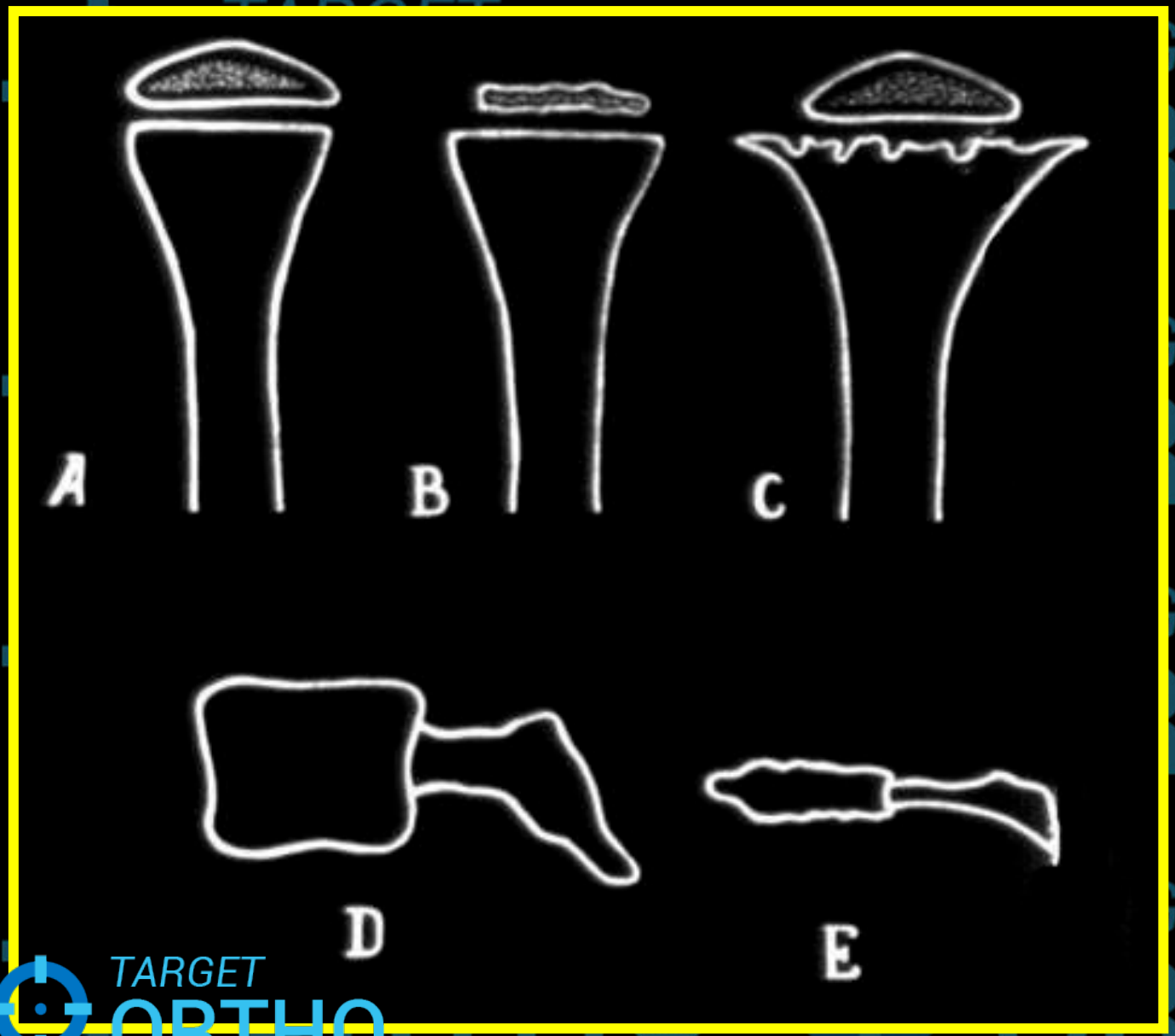
A. NORMAL

B. MULTIPLE EPIPHYSEAL DYSPLASIA TARDA



C. SPONDYLO-EPIPHYSEAL DYSPLASIA





INVOLVEMENT	DISEASE CATEGORY
A + D	NORMAL
B + D	EPIPHYSEAL DYSPLASIA
C + D	METAPHYSEAL DYSPLASIA
B + E	SPONDYLOEPIPHYSEAL DYSPLASIA
C + E	SPONDYLOMETAPHYSEAL DYSPLASIA

Nosology and classification of genetic skeletal disorders: 2023 revision.

SKELETAL DYSPLASIAS - DIAGNOSIS

FAMILY PEDIGREE

LAB STUDIES

PATHOLOGICAL STUDIES (BIOPSY,GENETIC MAPPING)

Any Skeletal Dysplasia

Epiphyseal involvement

Metaphyseal involvement

+/- Spine involvement

Bone Density

Miscellaneous

MULTIPLE EPIPHYSEAL DYSPLASIA

Epiphyseal involvement

Mild short stature

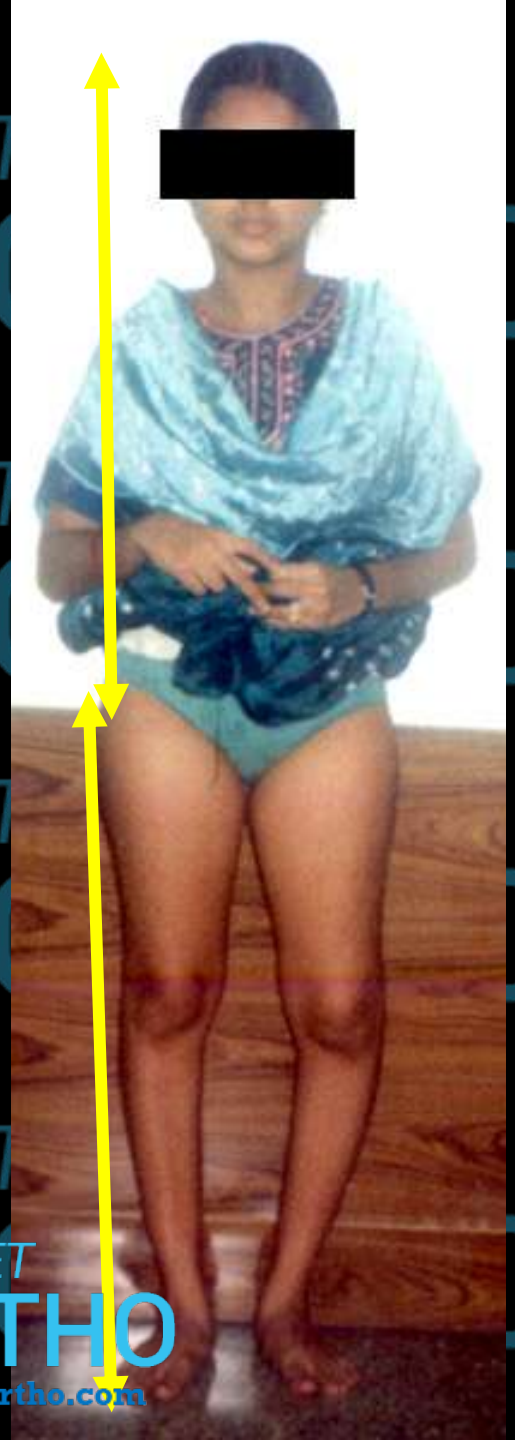
May have Pain

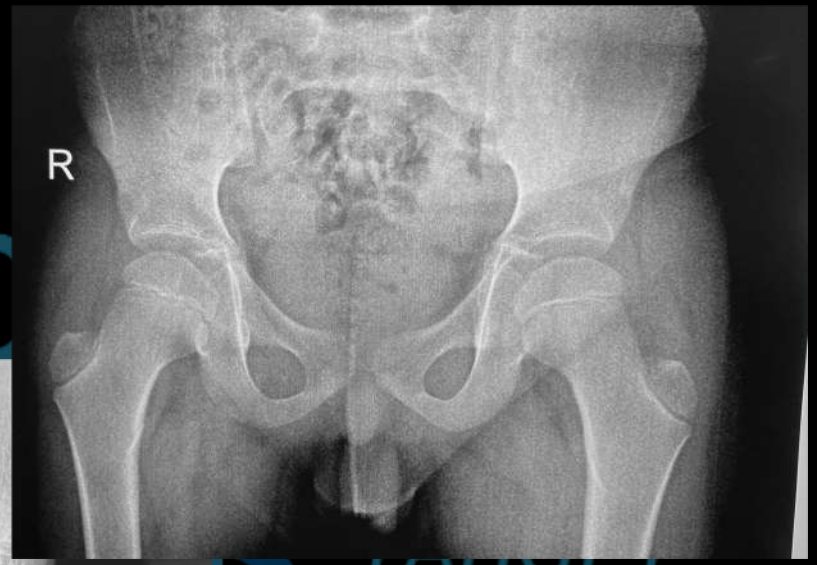
Symmetric ; Genu Varum, Coxa Vara

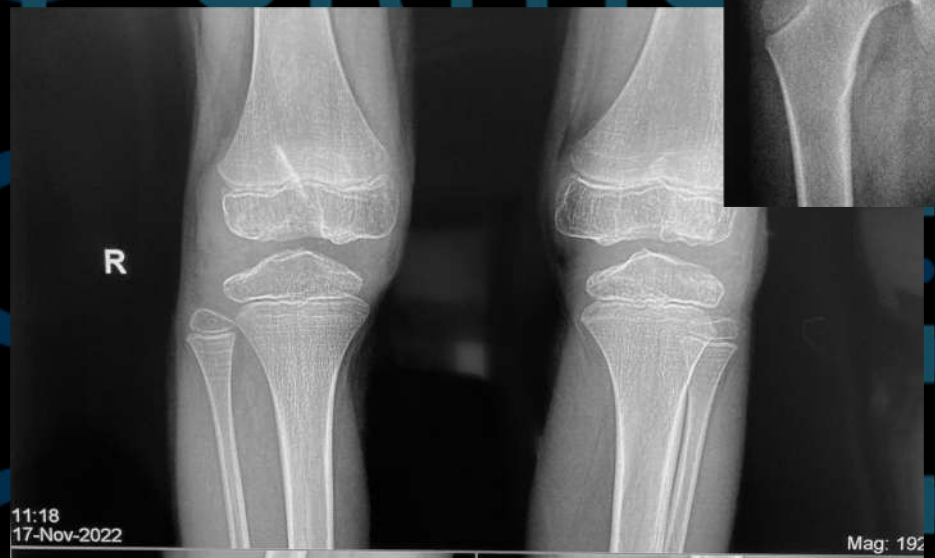
Few Surgery if any in childhood

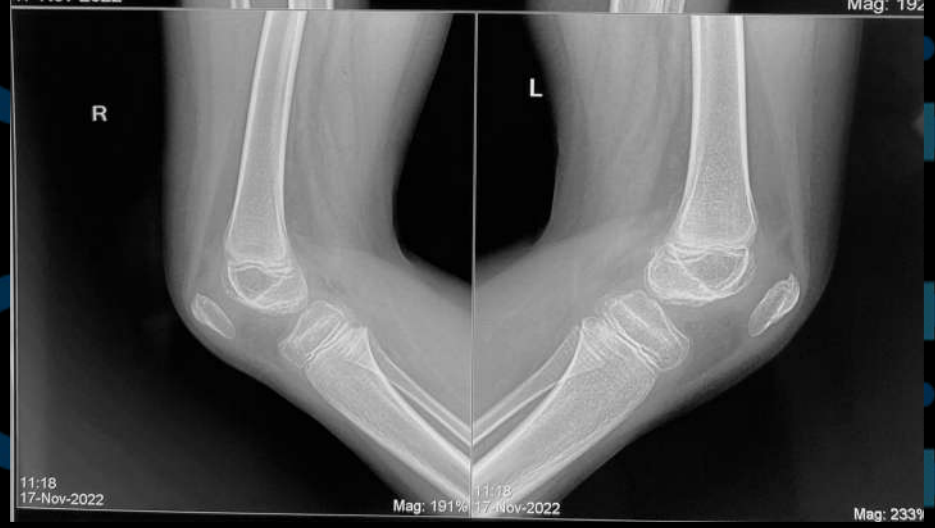
FACE , SKULL AND **SPINE - NORMAL**

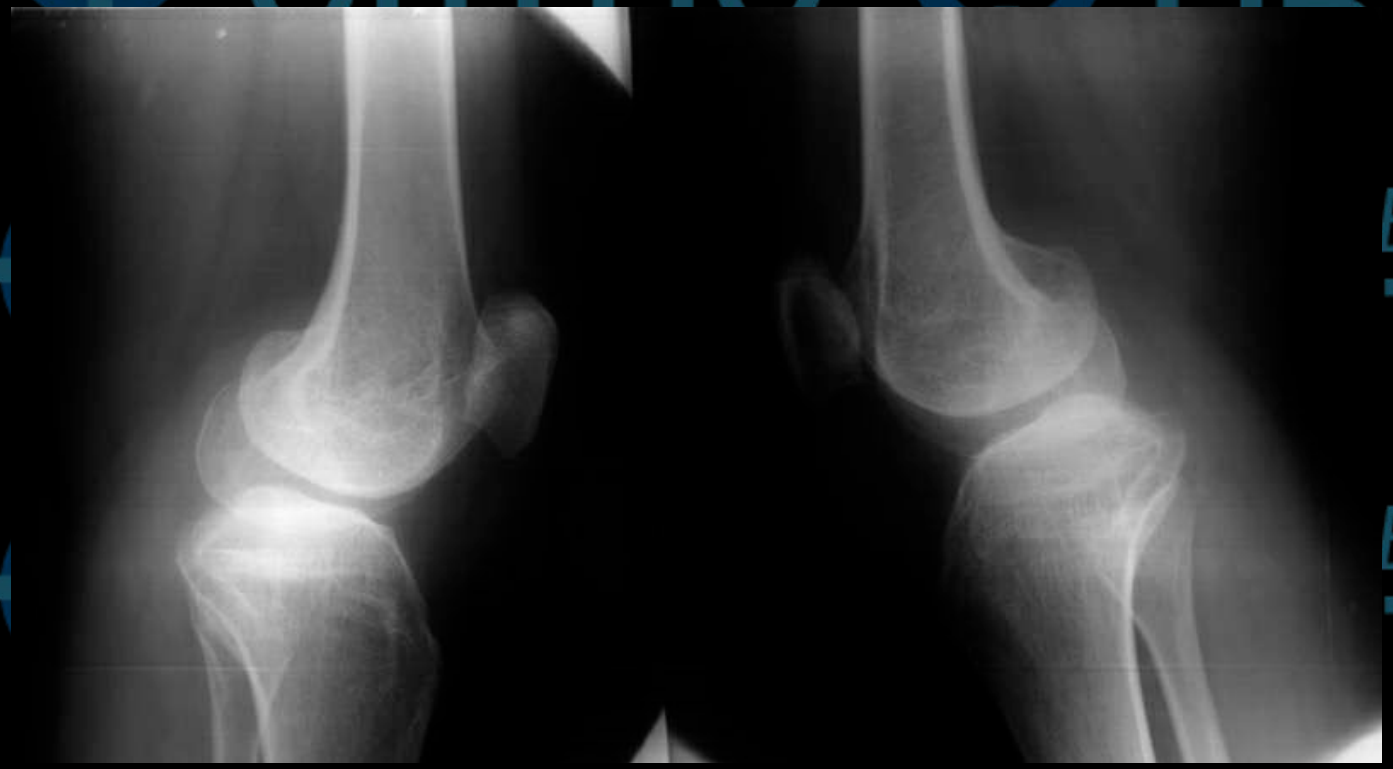
F 16 Yrs
(M.E.D)

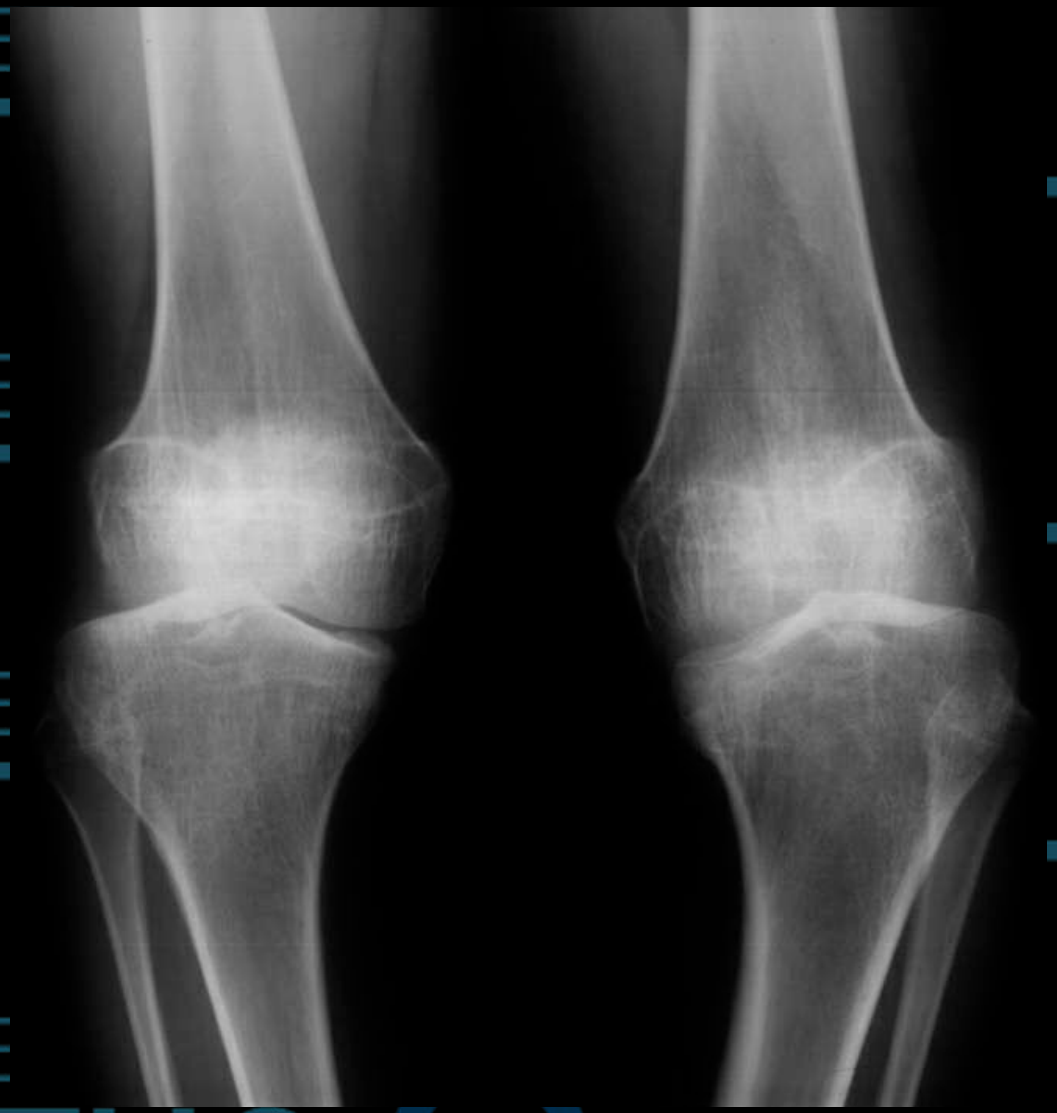




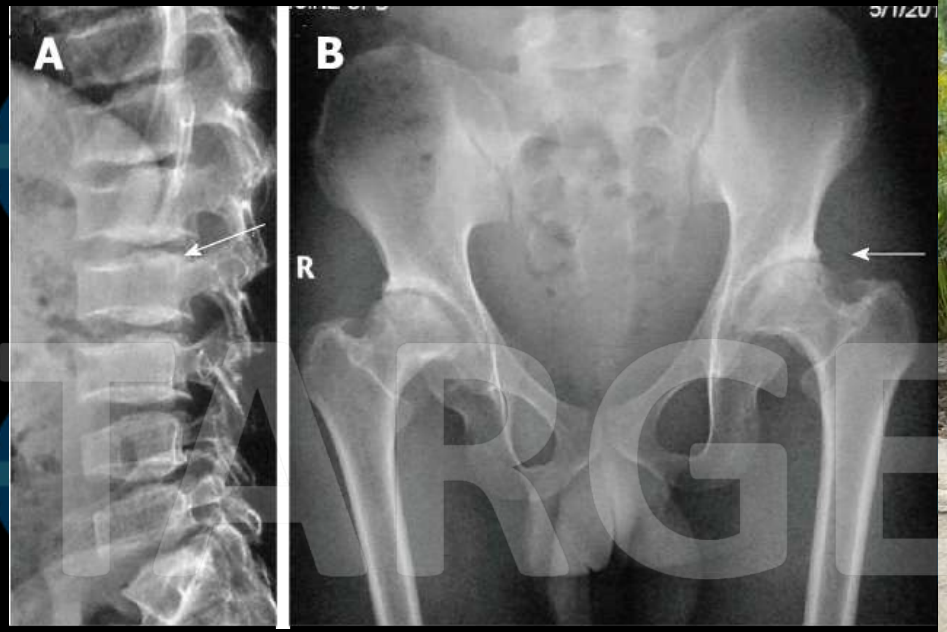








Spondyloepiphyseal Dysplasia



SED

- Short Trunk
- Short Stature
- AAI, CV Junction, Kyphosis
- Coxa Vara Waddling Gait
- Abnormal Type II Collagen

PSEUDOACHONDROPLASIA

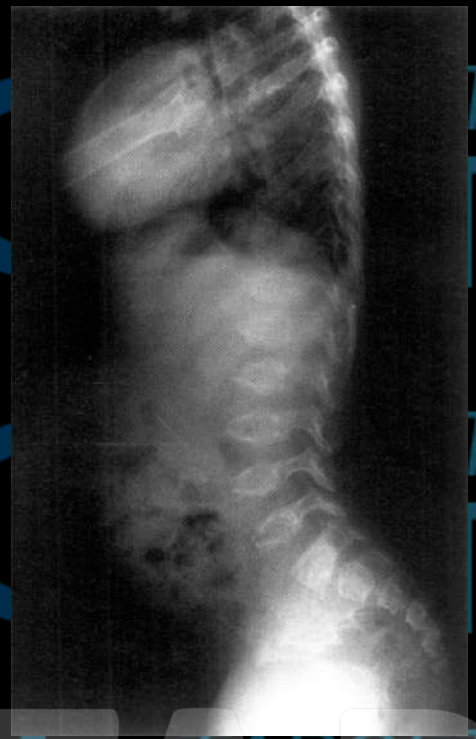
Epihyseal, Physeal And Metaphyseal Changes In Tubular Bones And Spine

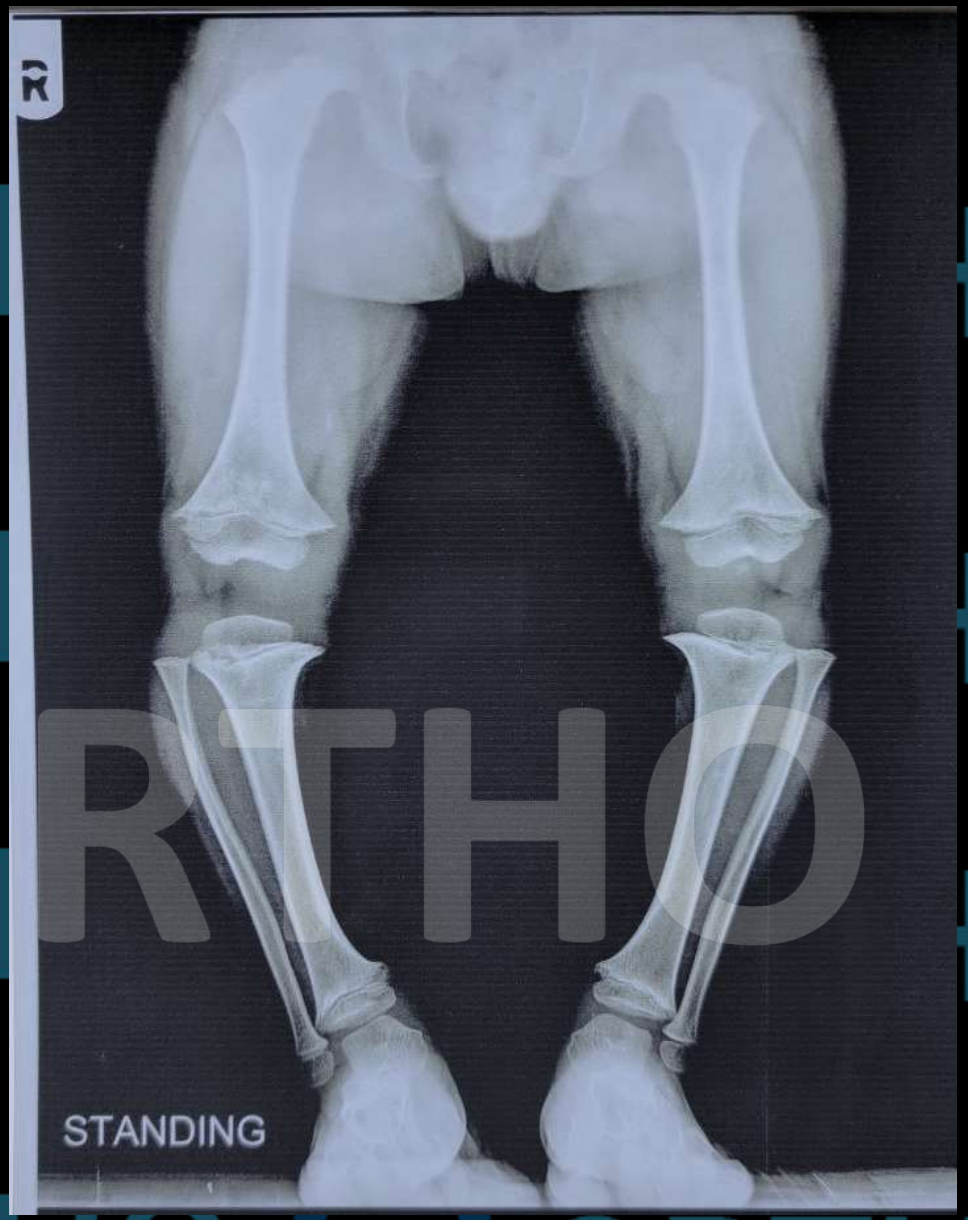
Short Limb Disproportionate Dwarfism

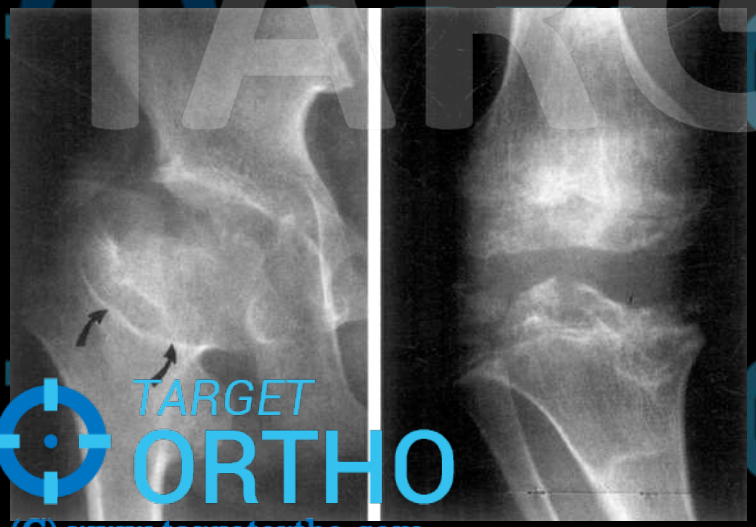
Head And Face Normal

Disease Manifests Between 2-3 Years Of Age

Adult Height – 3.5 – 4.5 Feet

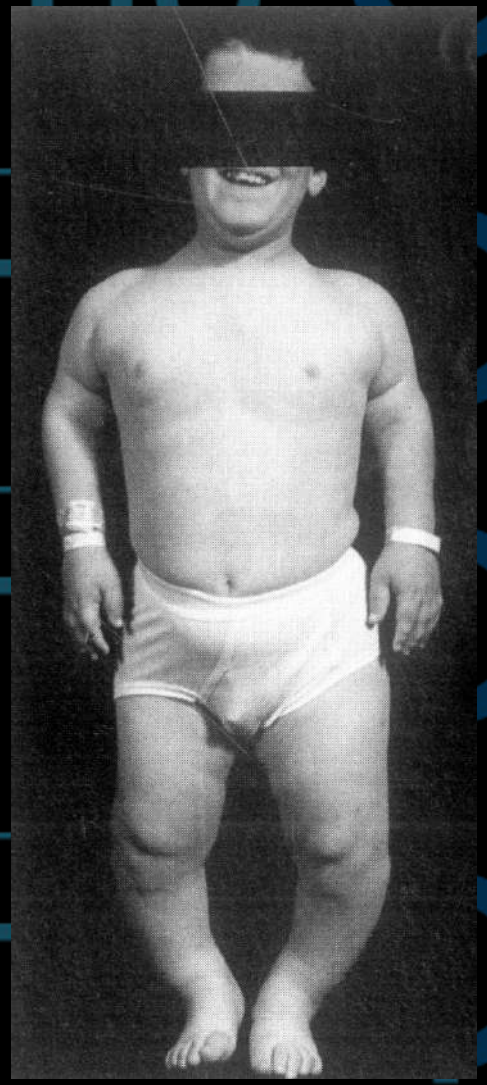


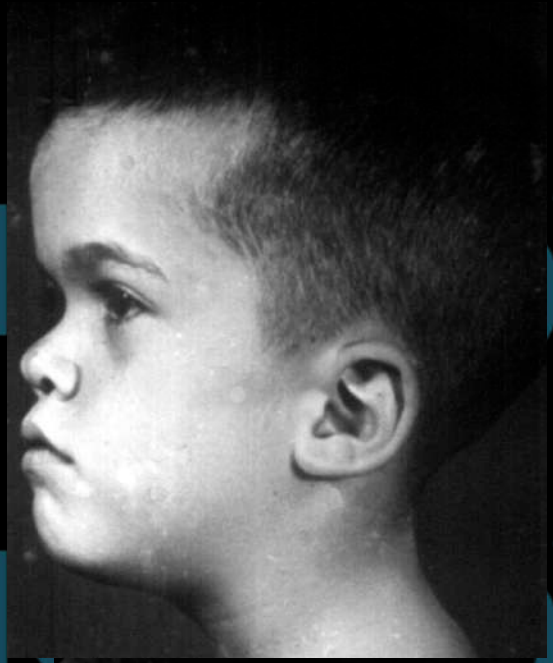


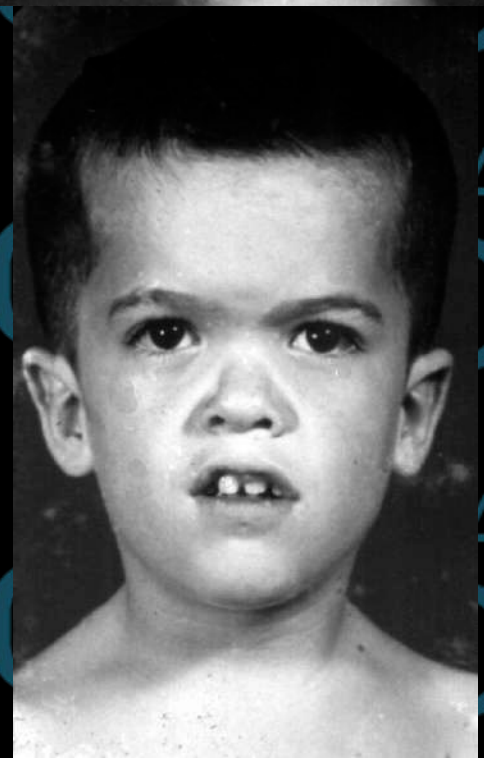


Epiphysis, metaphysis, Spine all involved

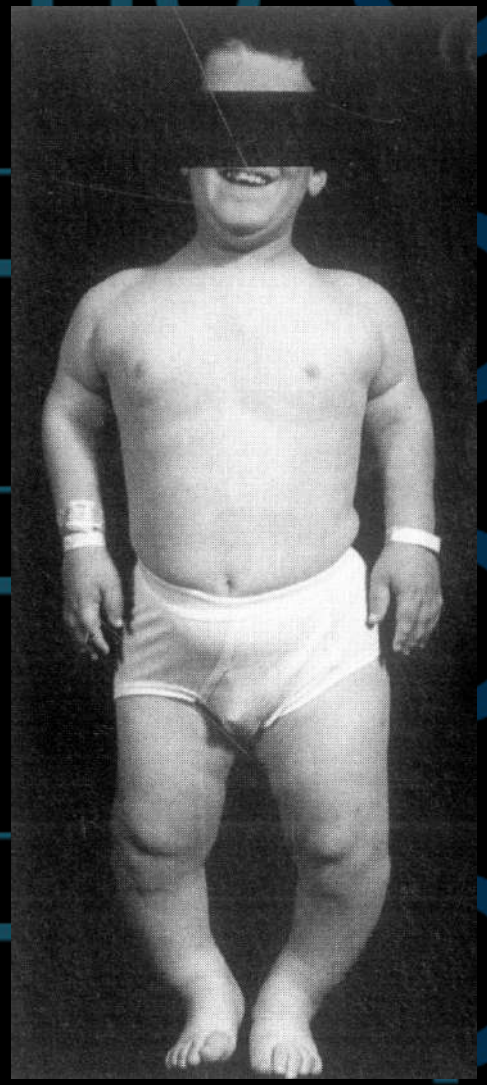
Achondroplasia

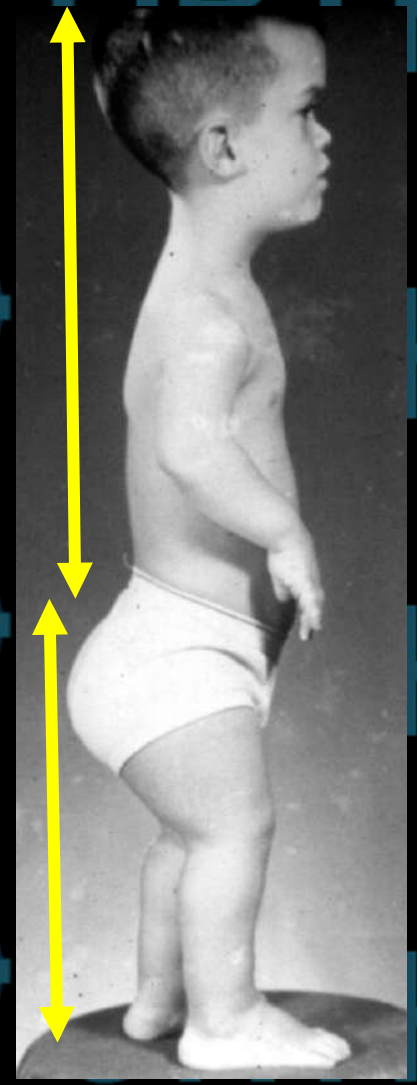


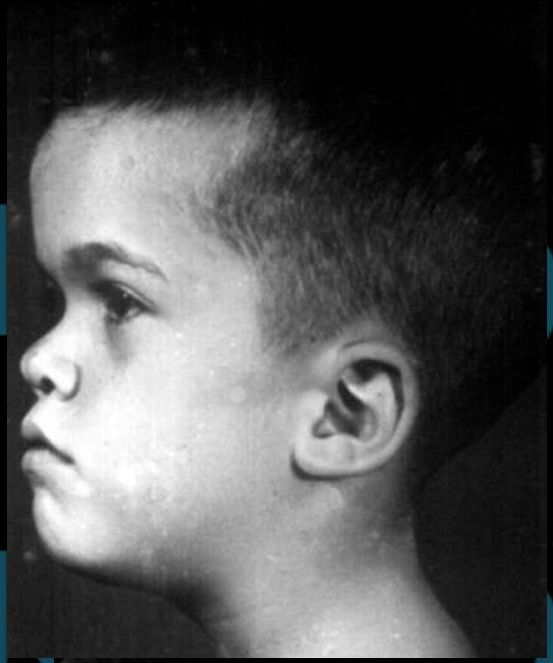


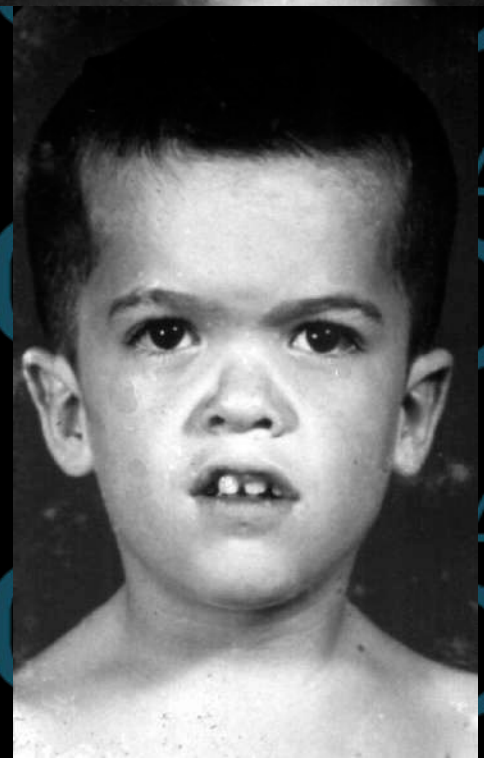


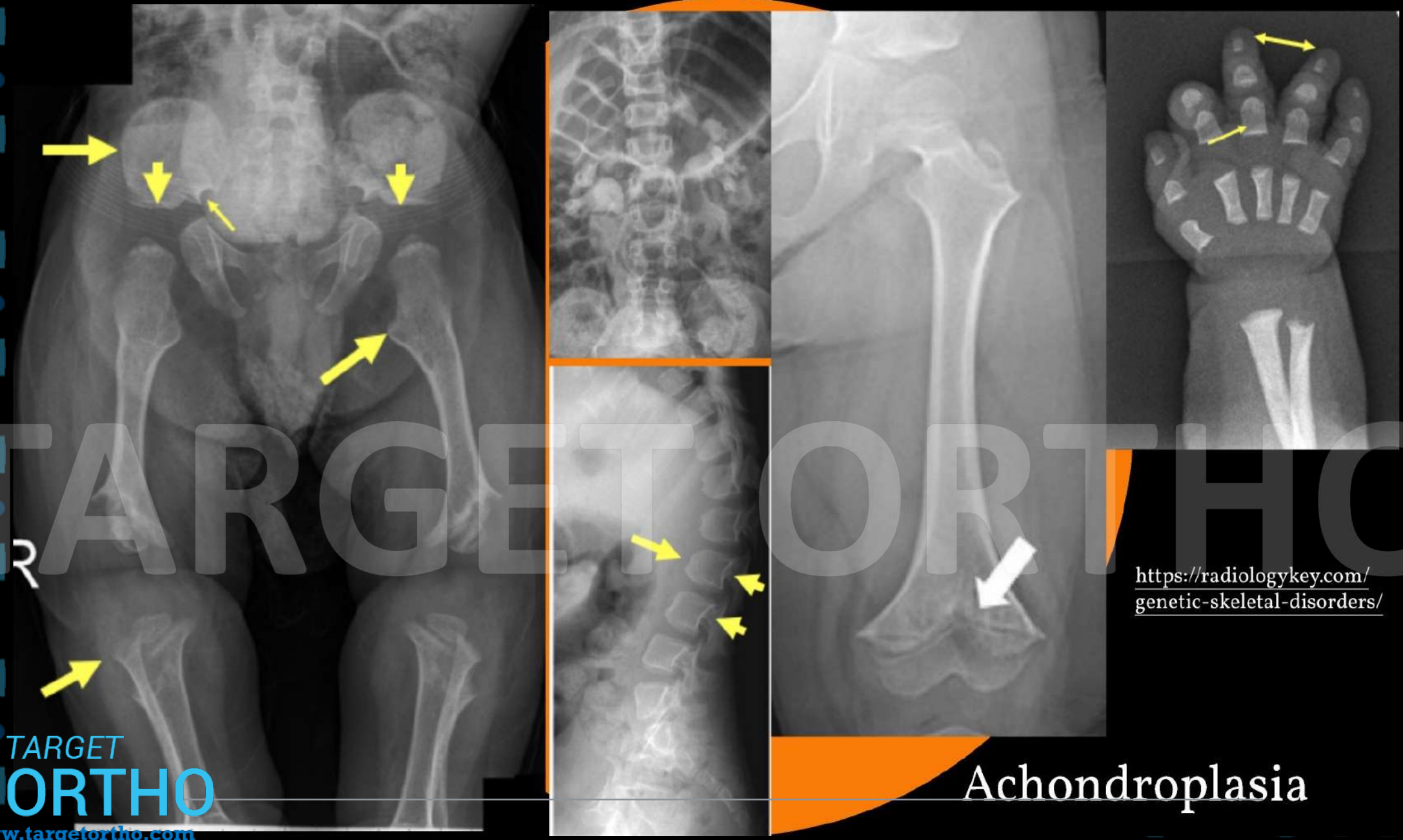
Achondroplasia





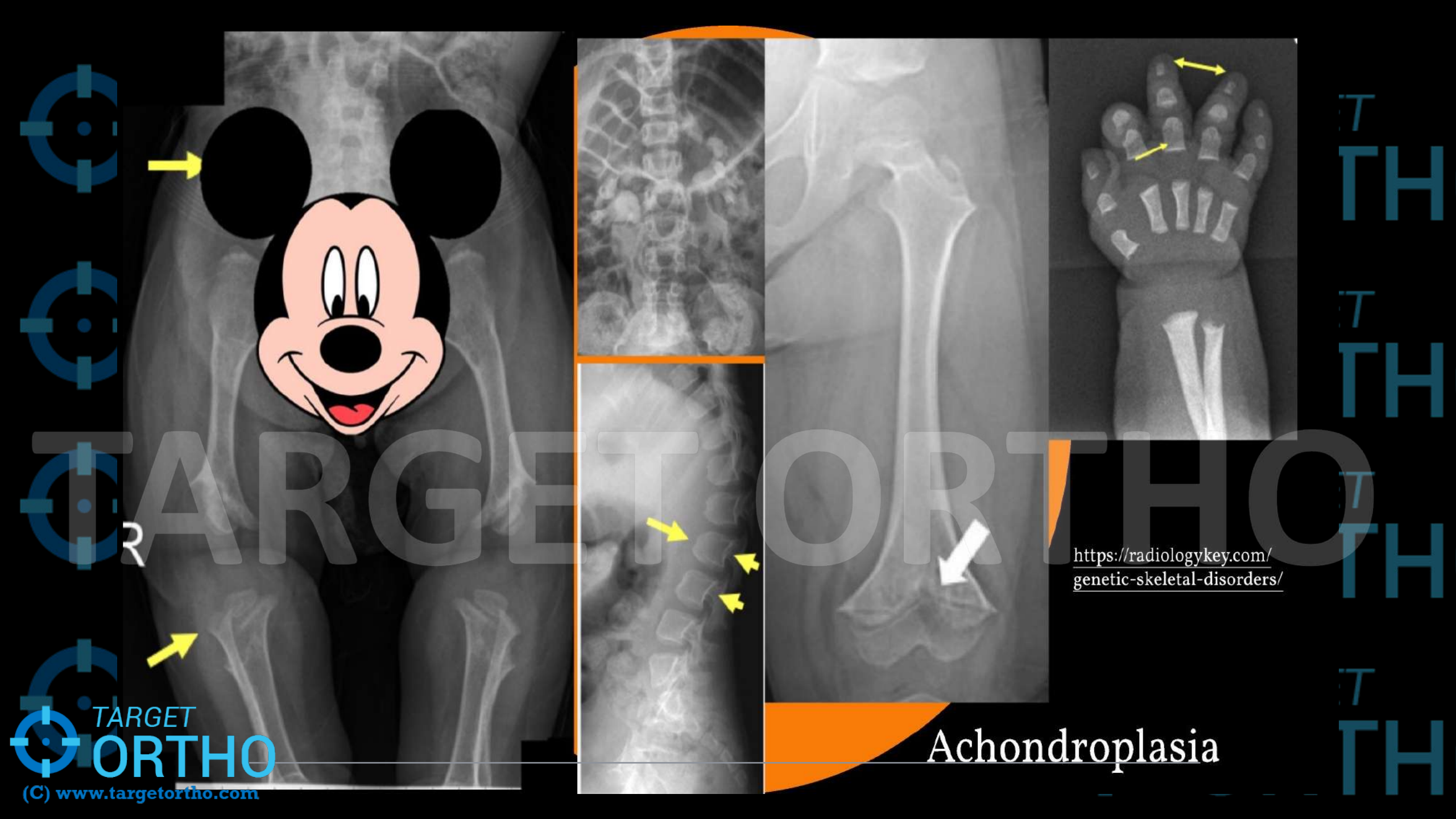




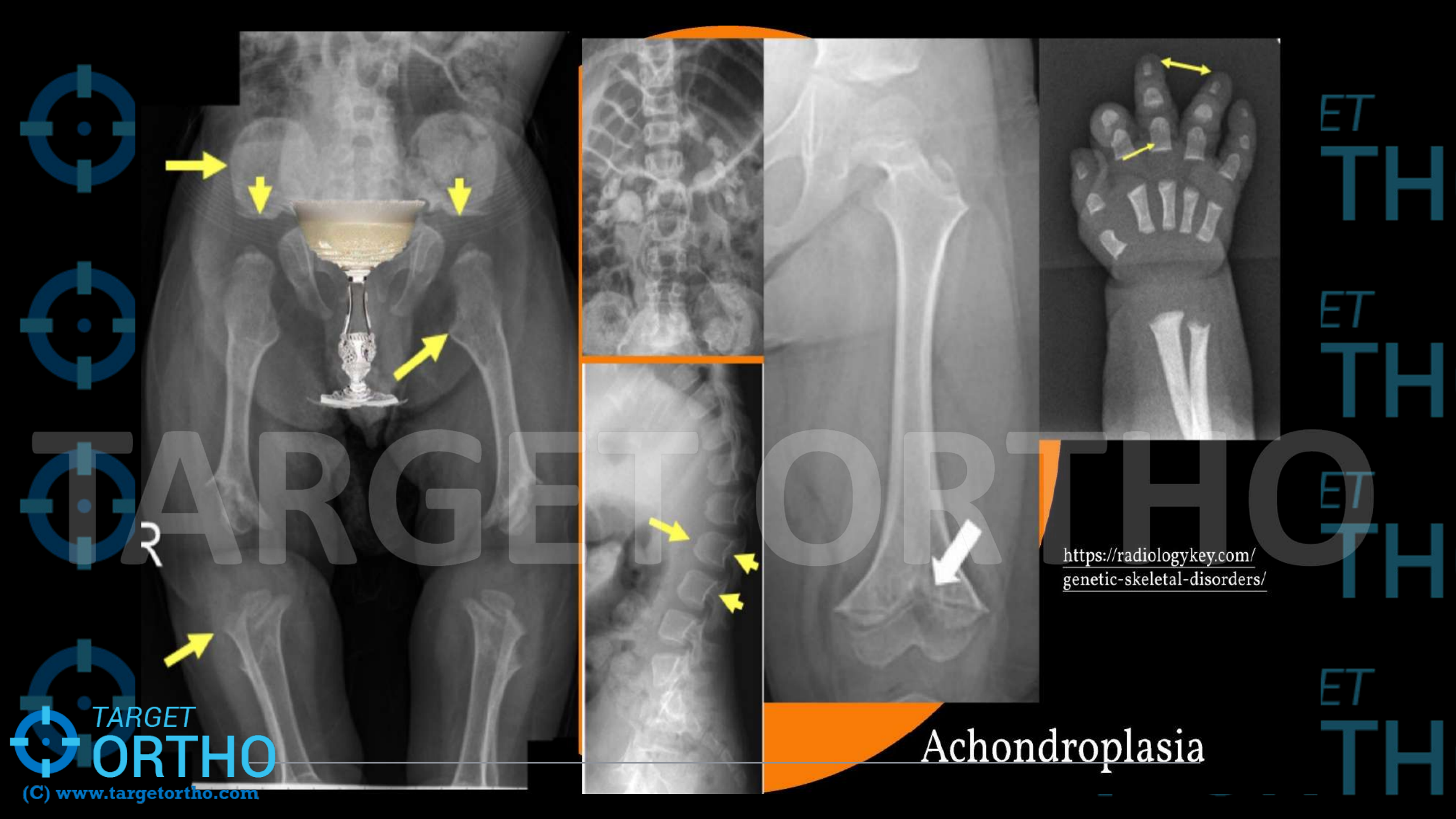


https://radiologykey.com/genetic-skeletal-disorders/

Achondroplasia



Achondroplasia



https://radiologykey.com/genetic-skeletal-disorders/

Achondroplasia

Bowing of the legs is the most common deformity in achondroplasia.

Brain base issue; AAI

Lumbar Canal stenosis (25%)

Koptis SE. Orthopedic aspects of achondroplasia: human achondroplasia in children. **Basic Life Sci** 1988

Differences between pseudoachondroplasia and achondroplasia

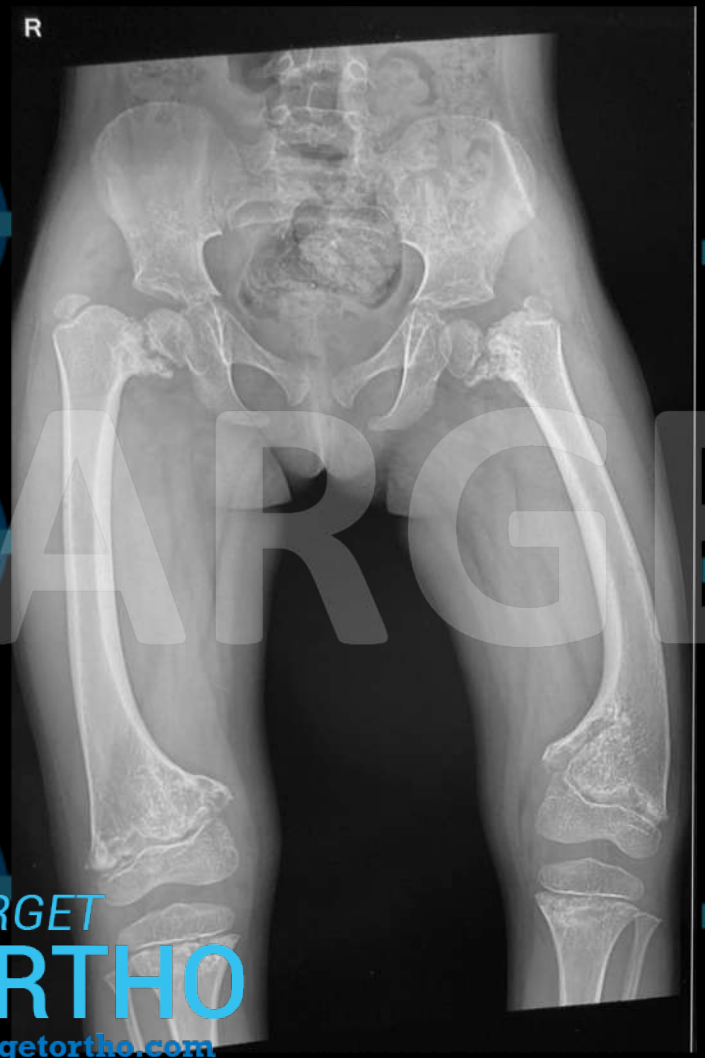
Pseudoachondroplasia	Achondroplasia	
Skull: Normal: "Achondroplasia with normal face"	Skull : Abnormal	
Spine: Platyspondyly +	Spine: Platyspondyly –	
Interpedicular distance normal	Interpedicular distance decreased in lumbar spine	
Epiphyses and metaphyses abnormal		Only metaphyses abnormal
Trident hand and champagne-glass pelvis absent	Trident hand and champagne-glass pelvis present	

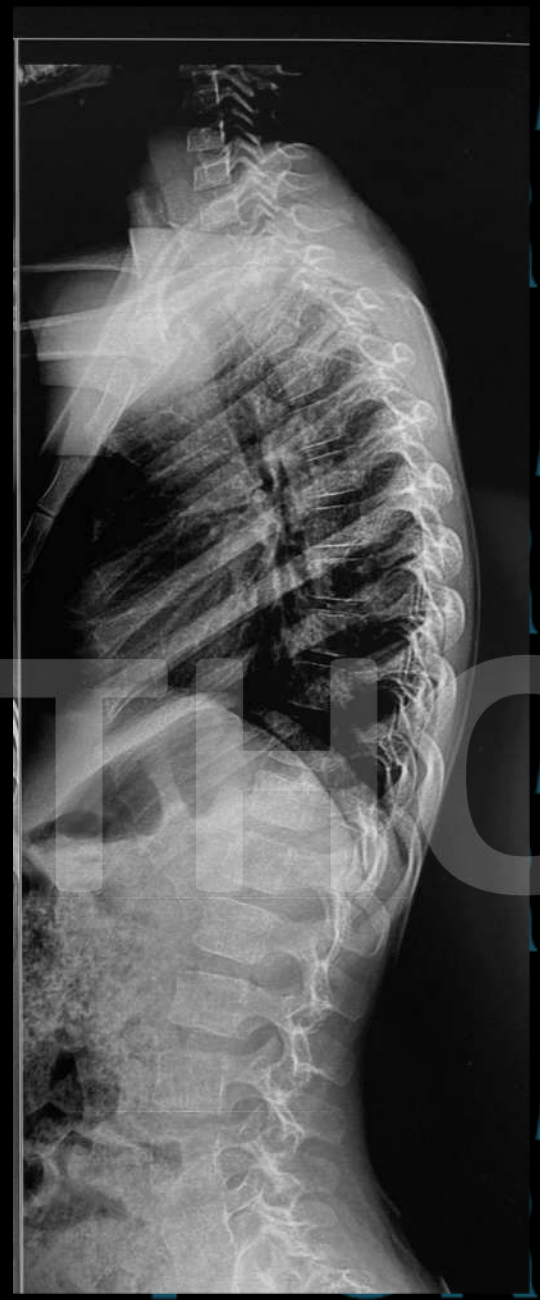
Metaphyseal Chondrodysplasia

Three main subtypes

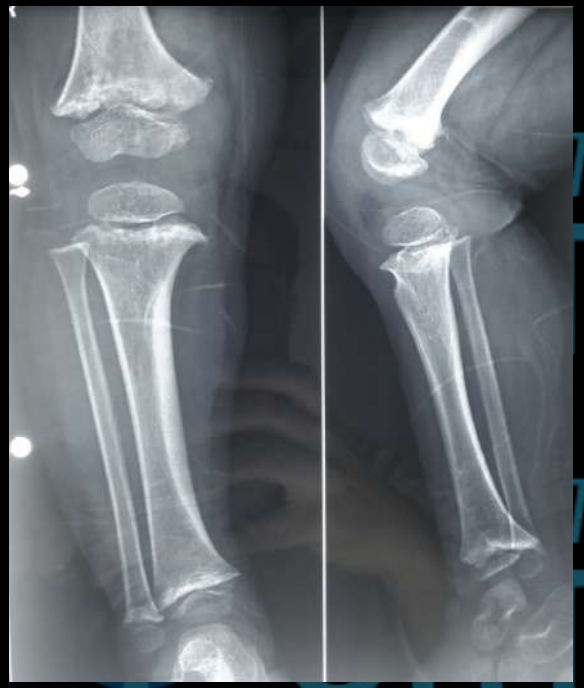
1. **Jansen (rare, most severe form)**
   autosomal dominant
   genetic defect in parathryoid hormone-related peptide (PTHrP)
2. **Schmid (more common, less severe form)**
   autosomal dominant
   Genetic defect in type X collagen
3. **McKusick**
   autosomal recessive
   most commonly occurs in Amish and Finnish populations
   cartilage-hair dysplasia (hypoplasia of cartilage and small diameter of hair)

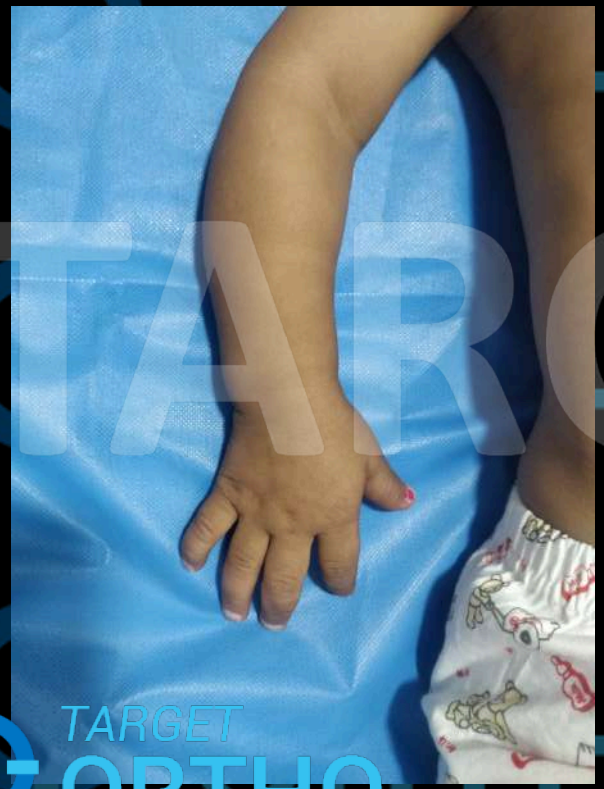
Metaphyseal Dys

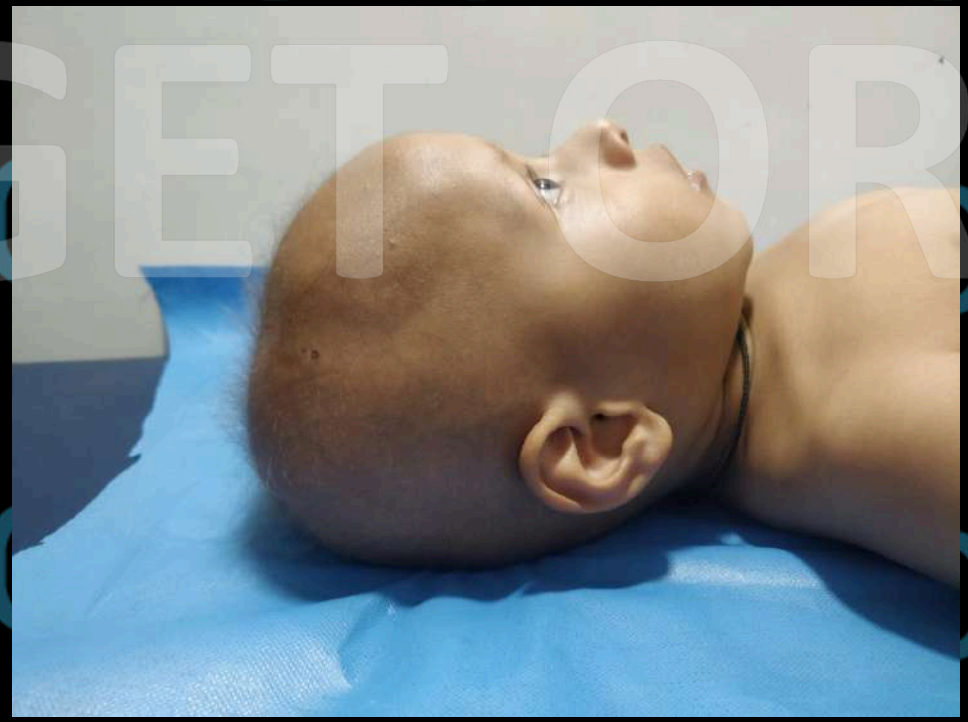


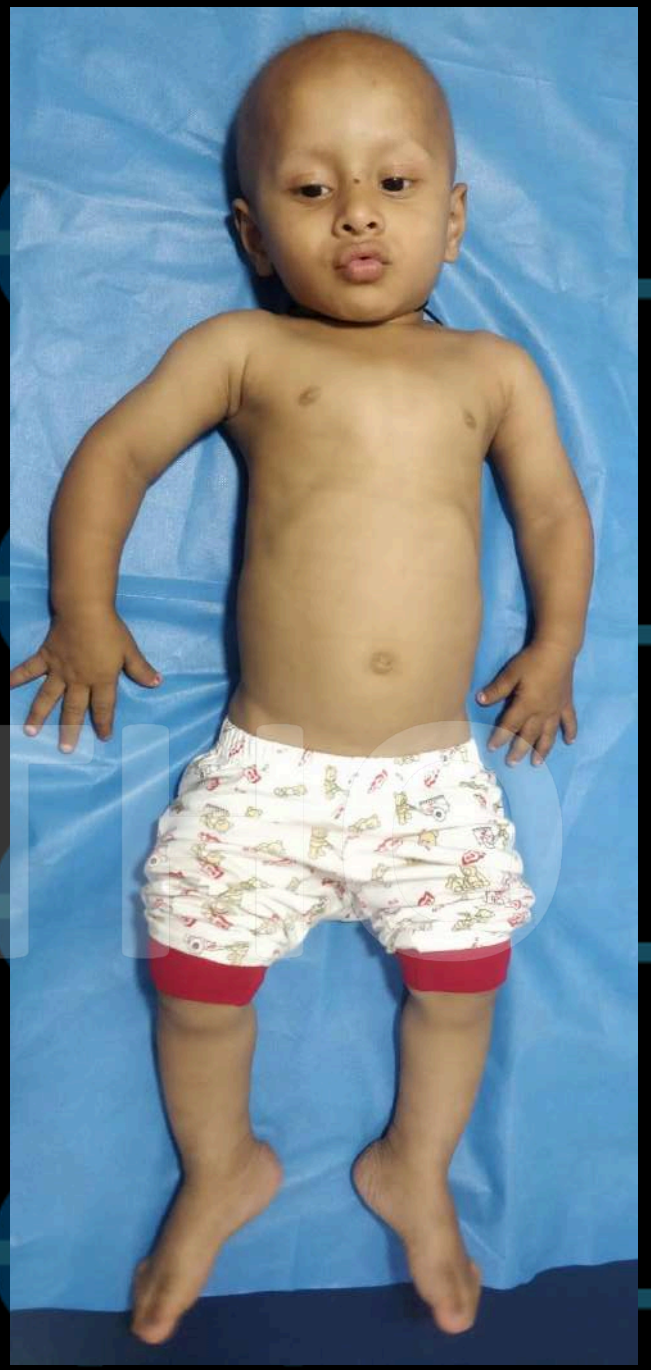


McCusick syndrome









Any Skeletal Dysplasia

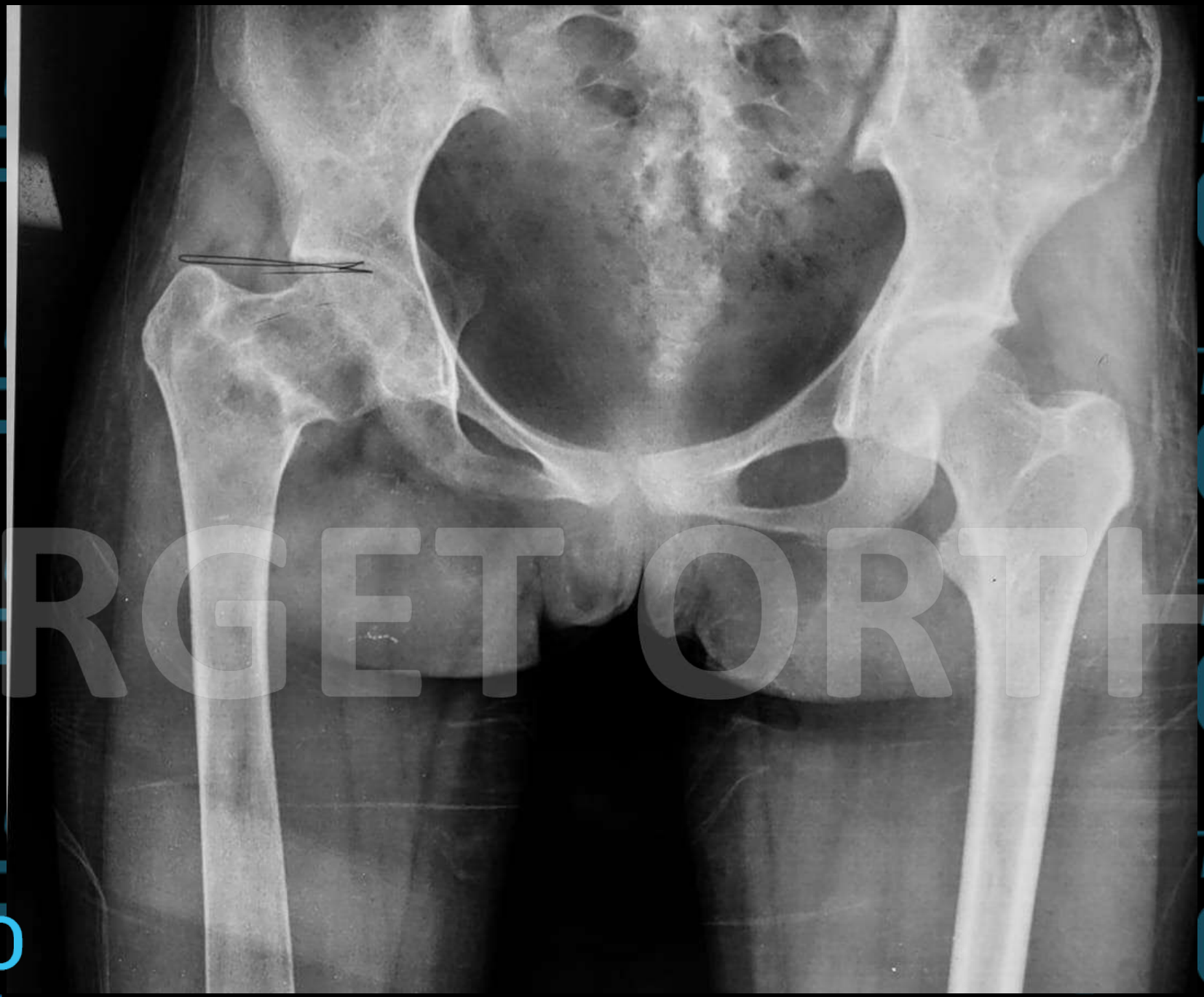
Epiphyseal involvement

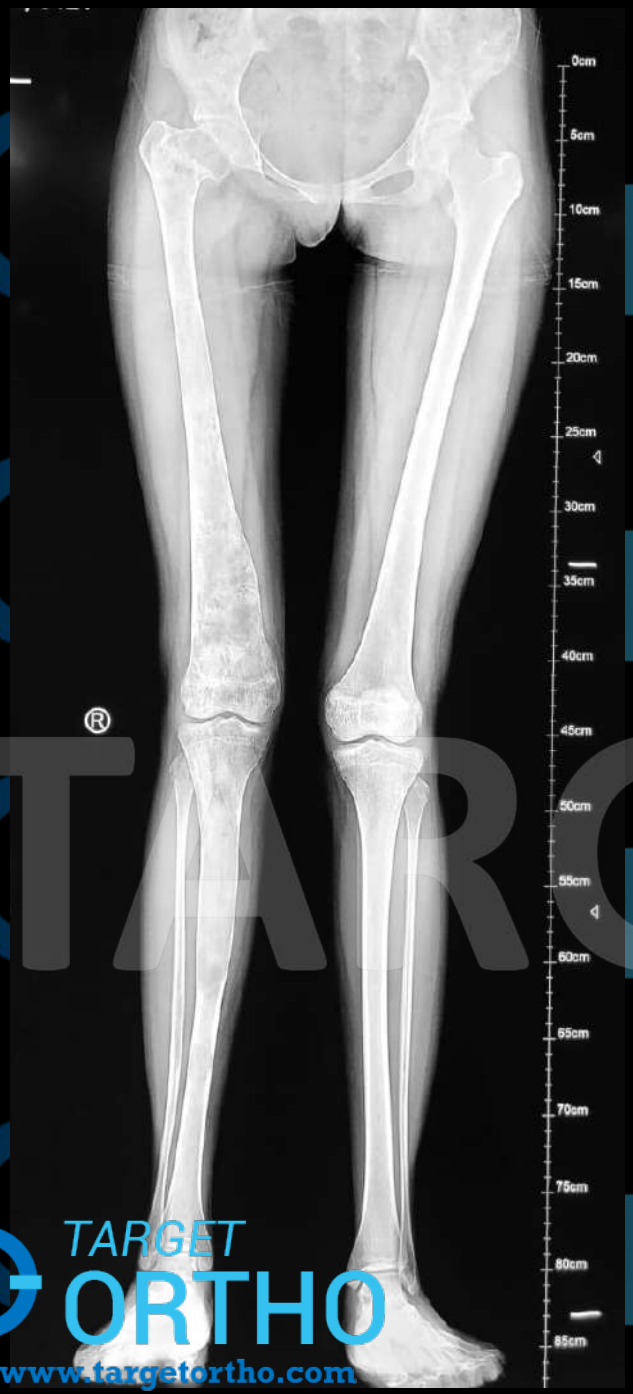
Metaphyseal involvement

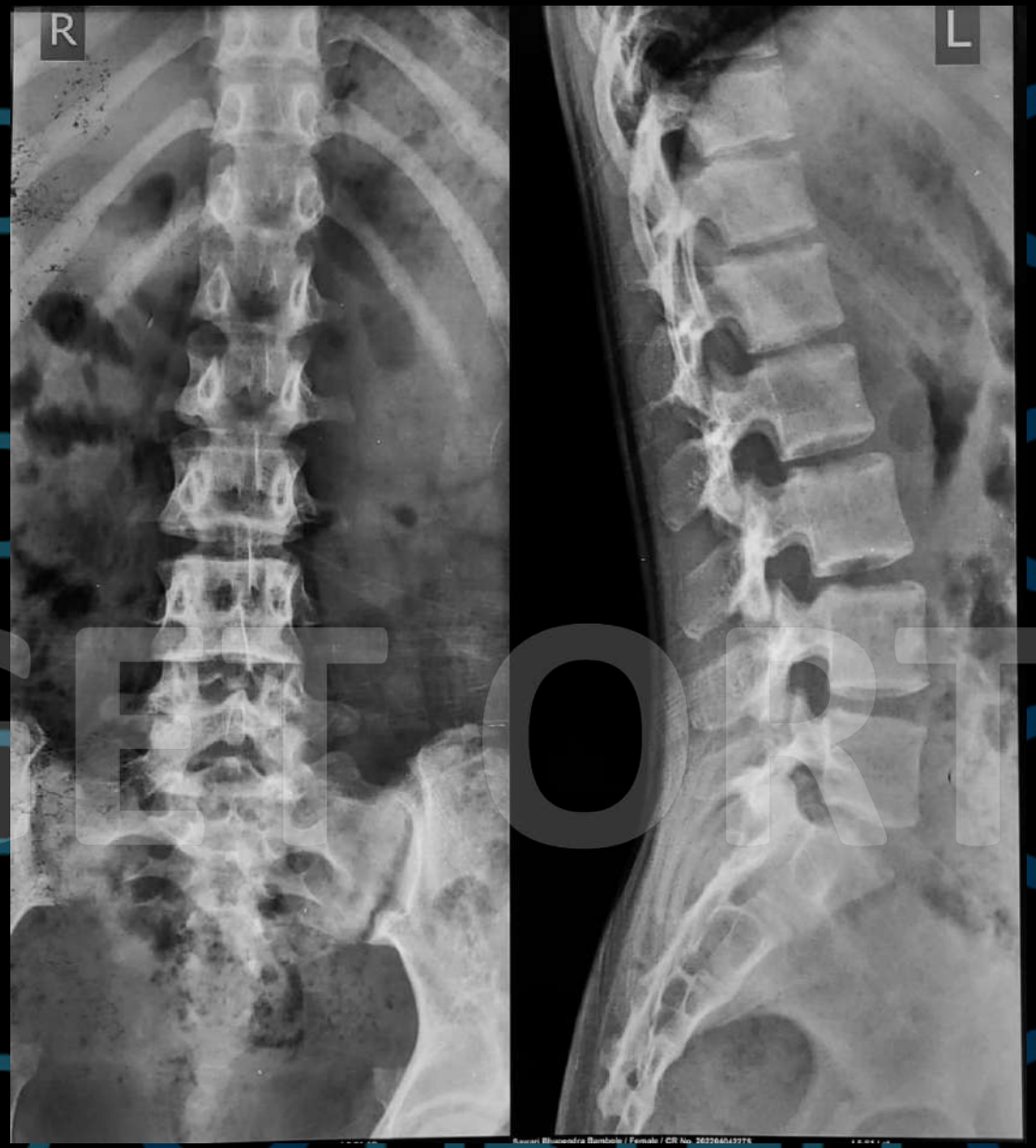
+/- Spine involvement

Miscellaneous

Bone Density





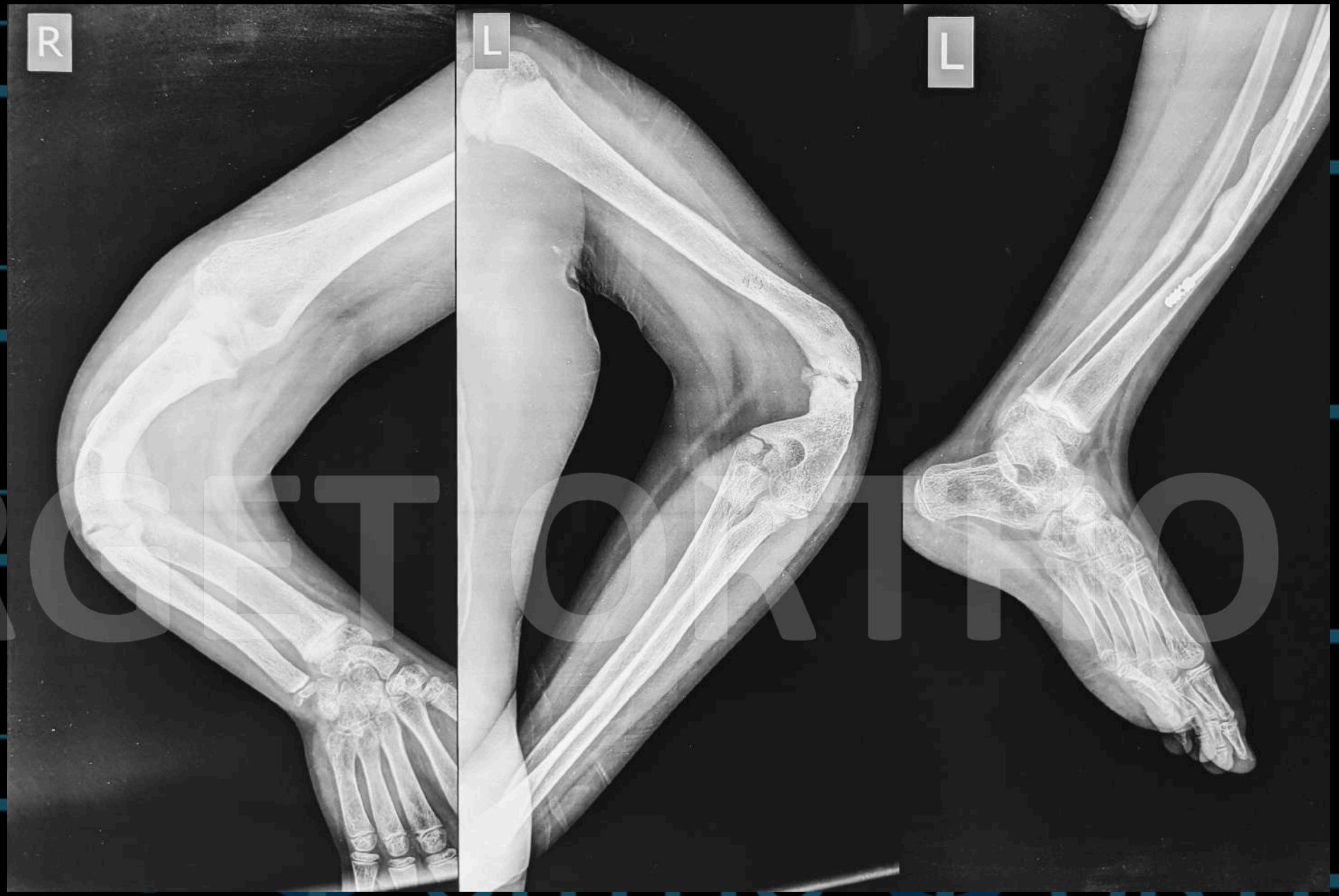


Multiple Lytic lesion

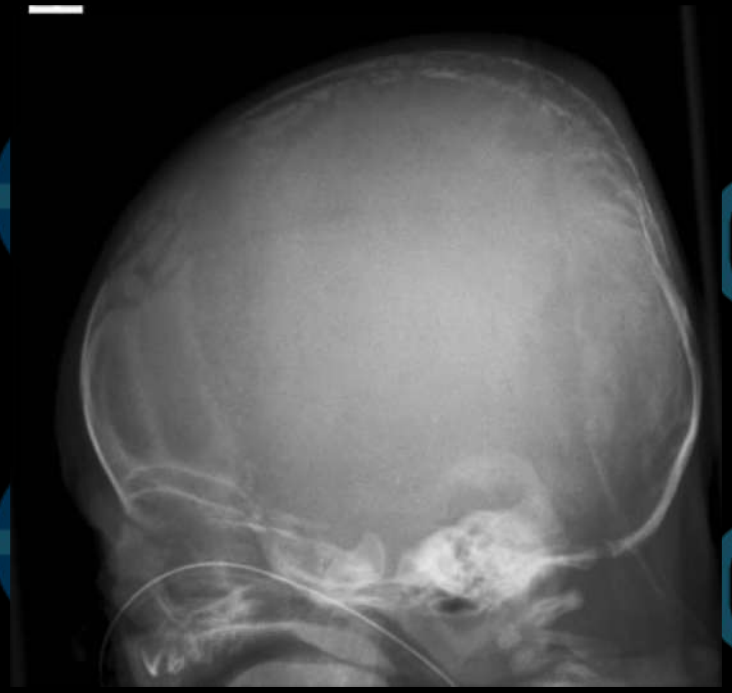
Fibrous matrix

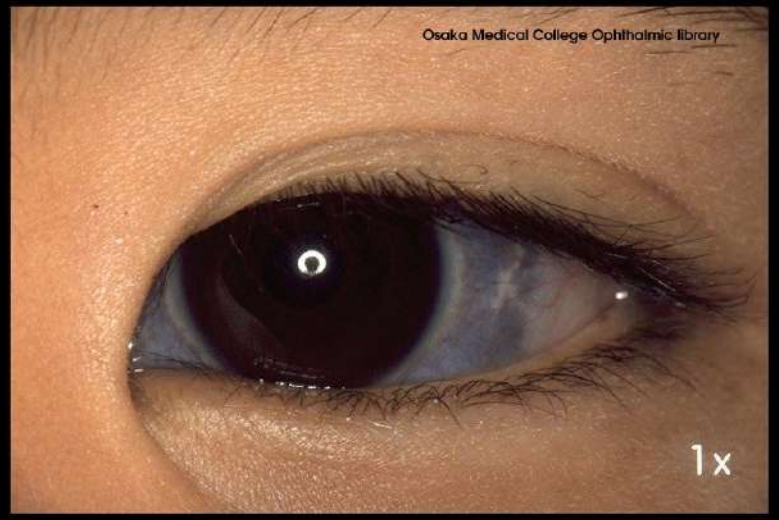
Diagnosis:
Fibrous dysplasia

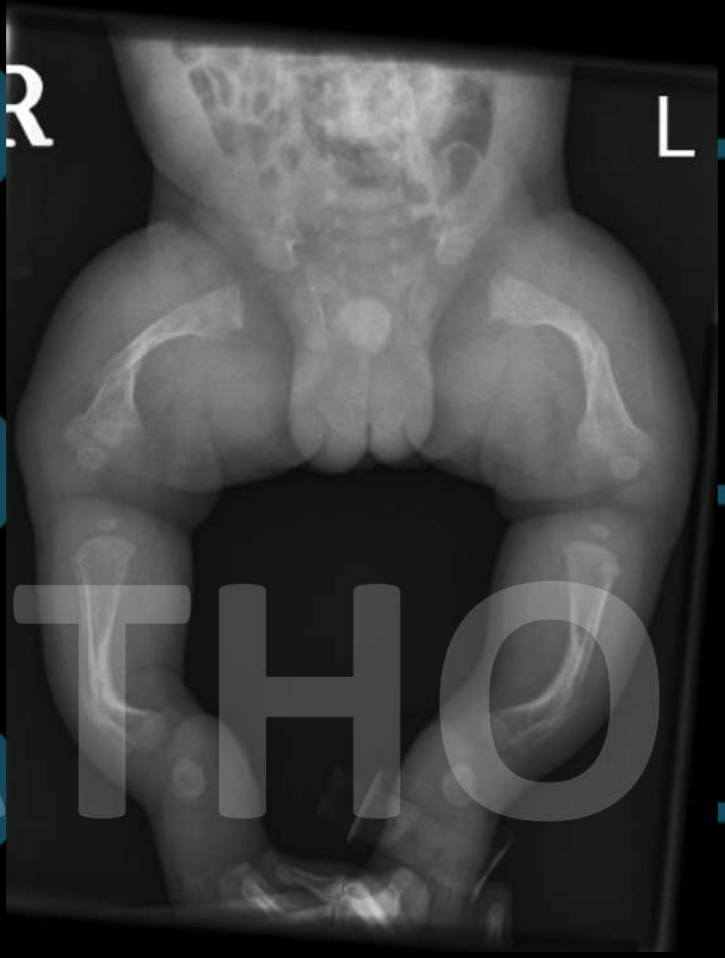
Osteogenesis Imperfecta

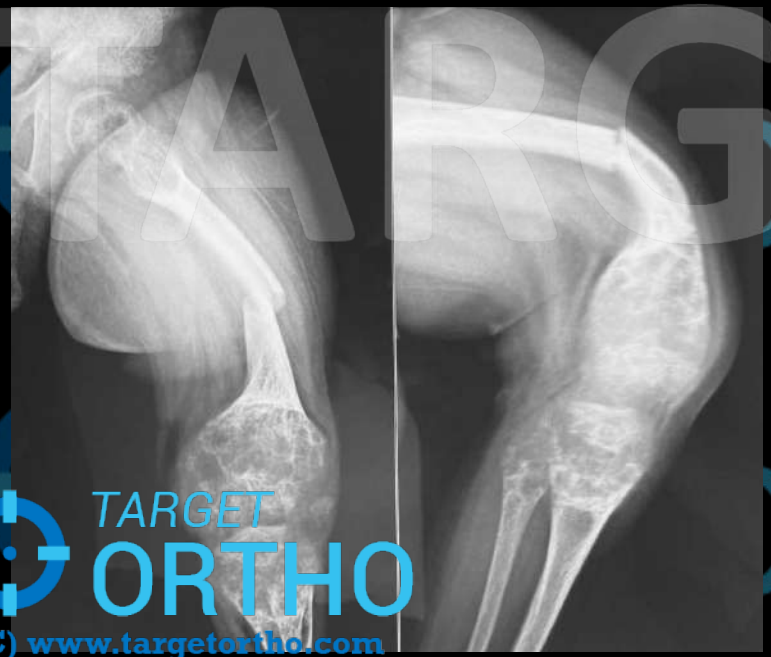


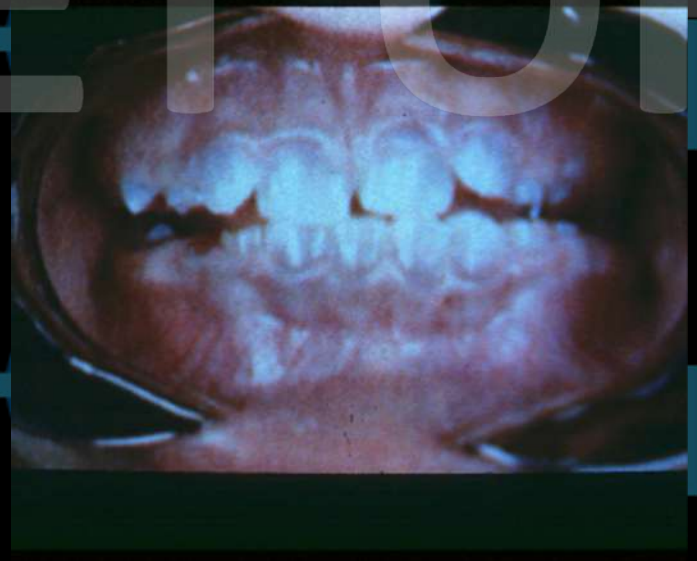
COL 1A1 Mutation- OGI











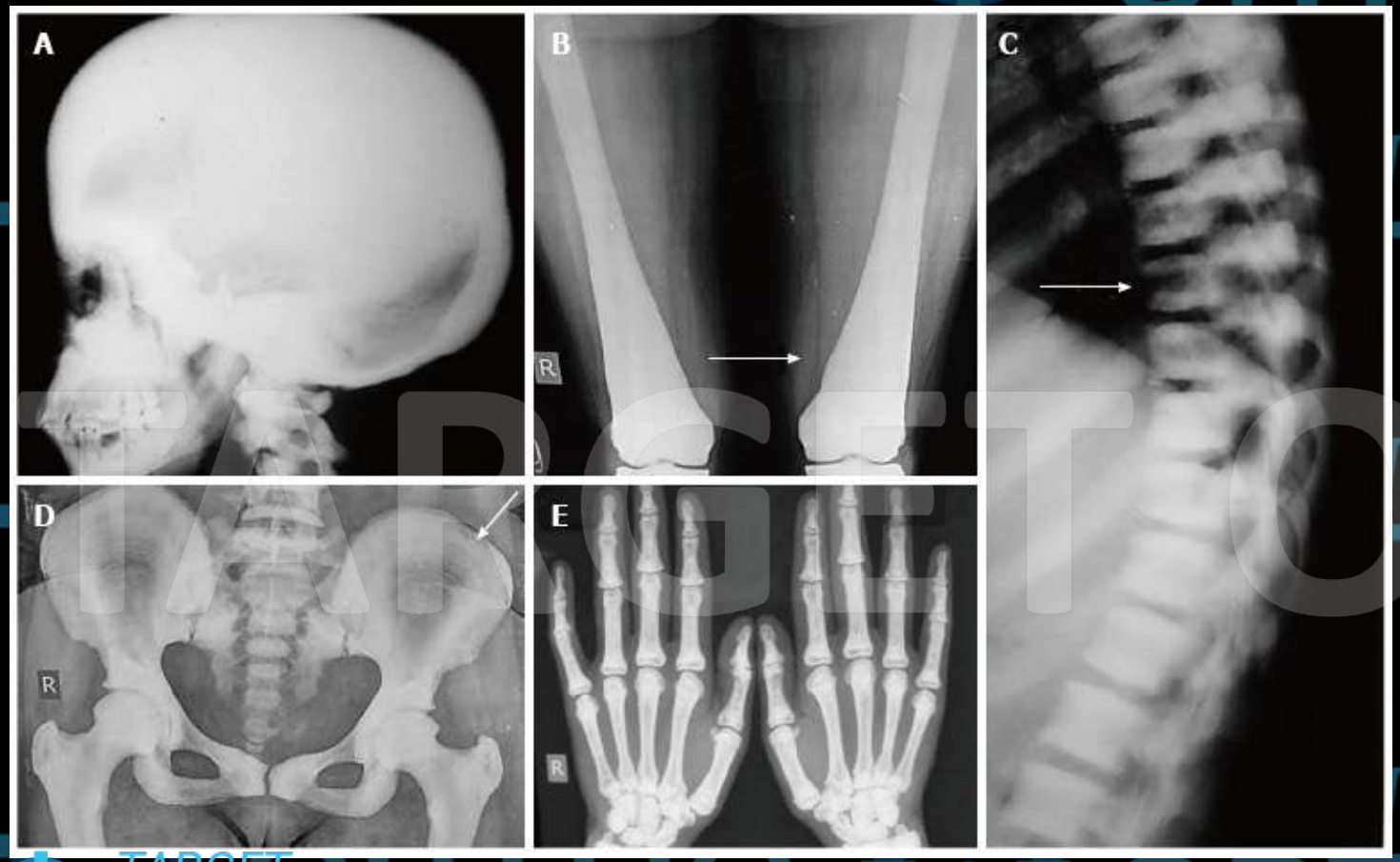
Sclerosing bone dysplasia

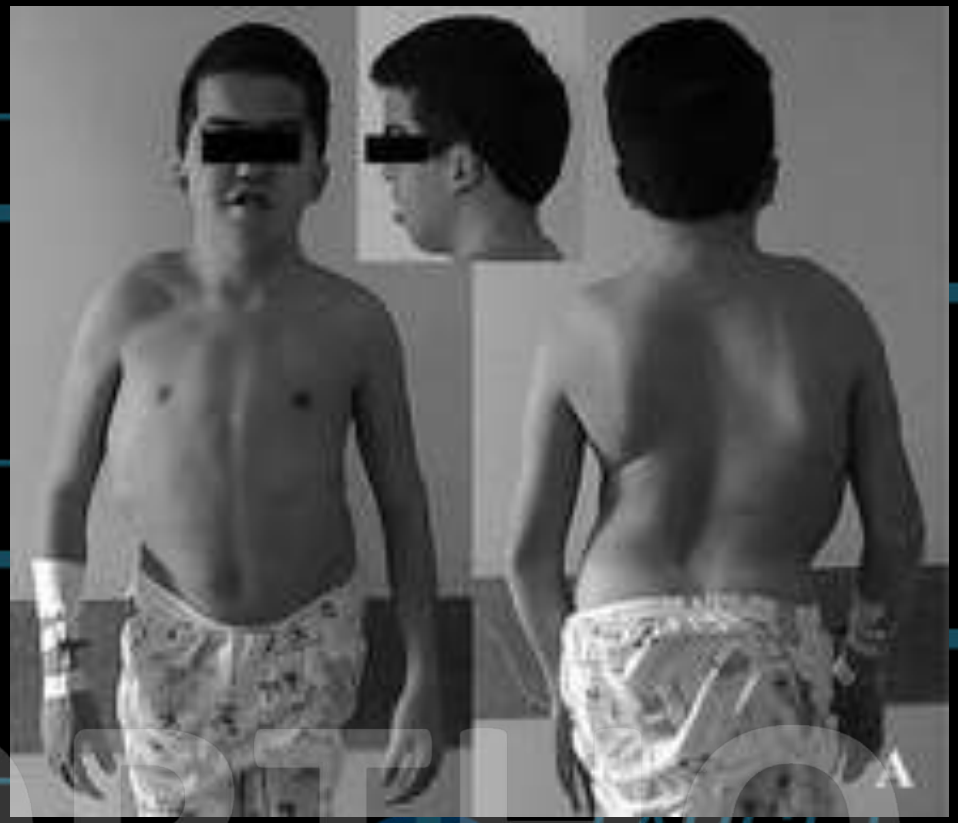
- **Enchondral Bone issue**
  Osteopetrosis
  Osteopoikilosis
  Pyknodysosotosis

- **Intramembranous bone issue**
  Progressive diaphyseal dysplasia

- **Mixed sclerosing dysplasias**
  Melorheostosis and overlap syndromes

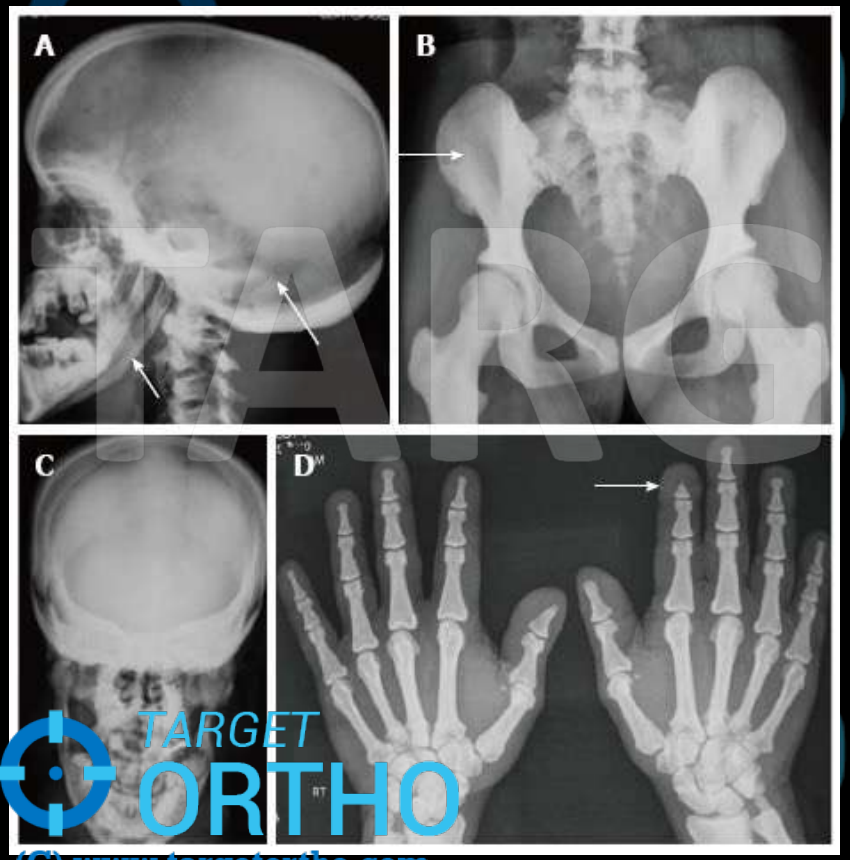
Osteopetrosis

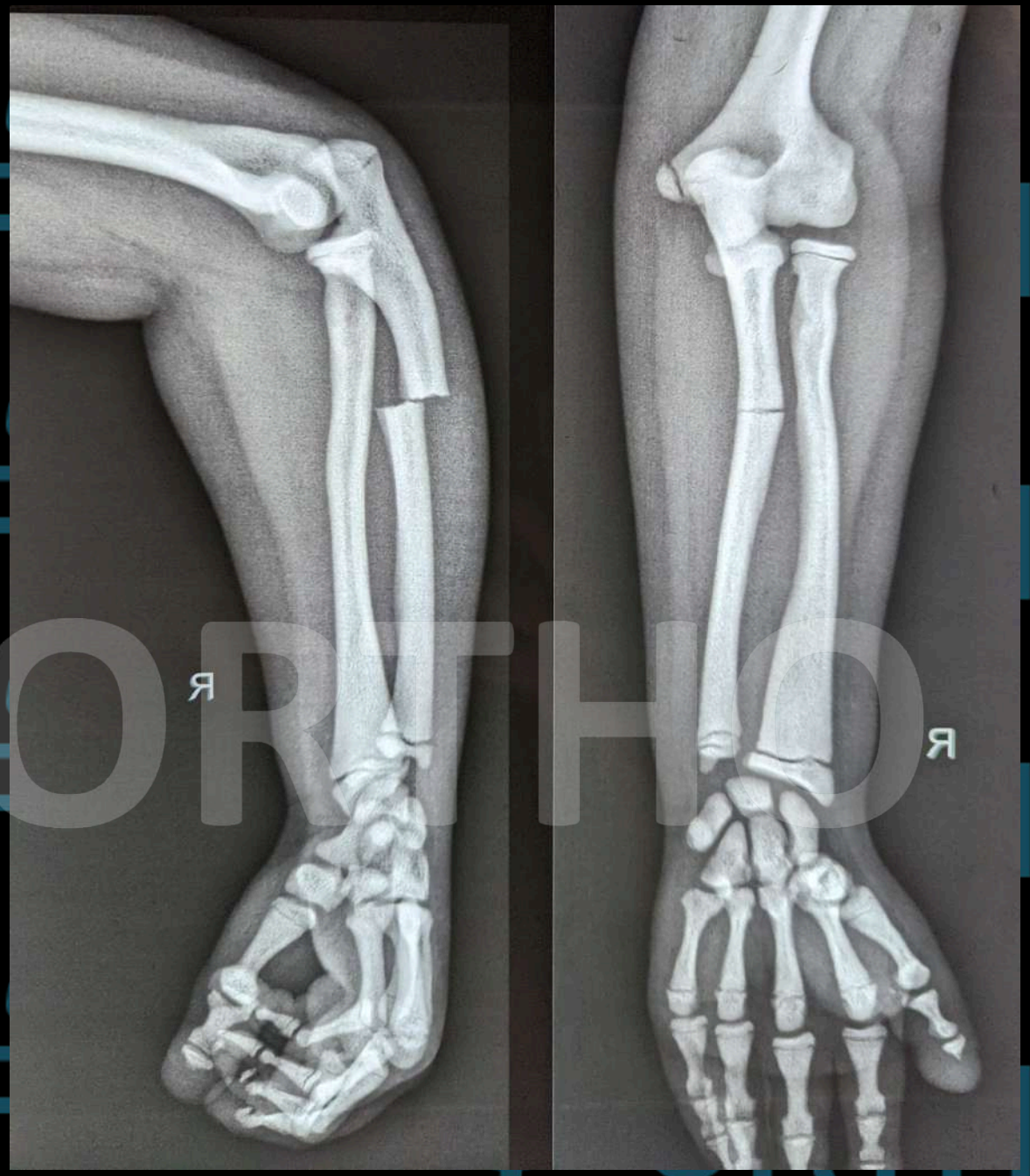




Erlenmeyer flask
Skull, Femur, hand Increased density
Prognathism
Spine, Bone in Bone

Pyknodysostosis





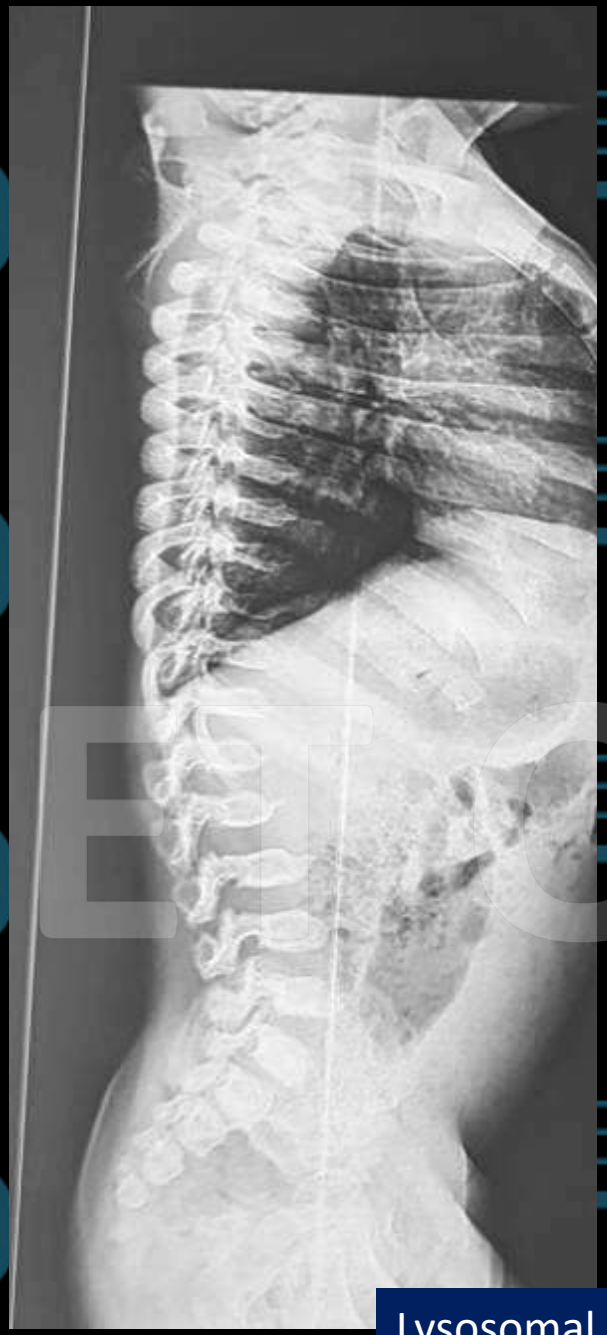
Mucopolysaccharidoses

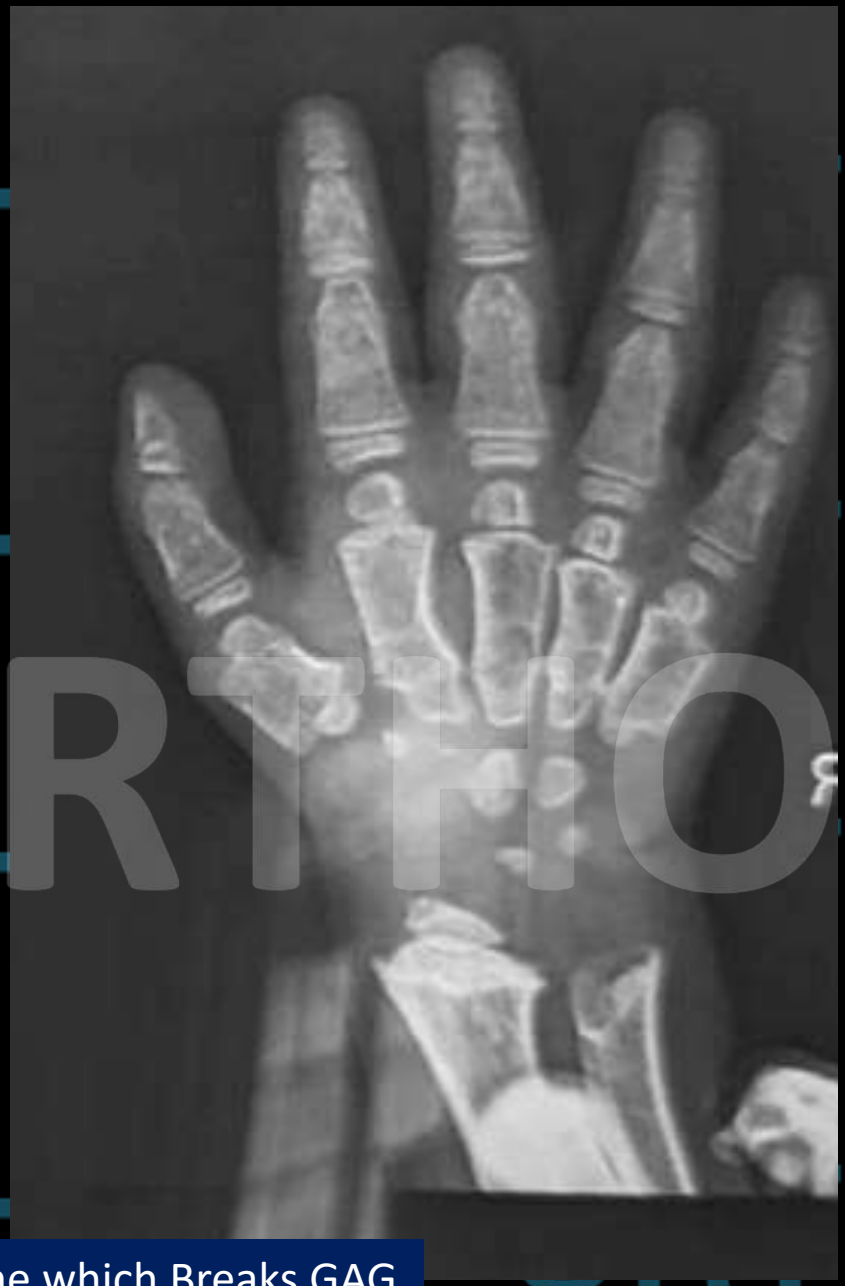
	Morquio	Hurler	San Filippo	Hunter
Pathophysiology	Type A (galactosamine-6-sulfate-sulphatase deficiency). Type B (beta-galactosidase deficiency)	Caused by alpha-L iduronidase deficiency	Multiple enzyme deficiencies	Sulpho-iduronate-sulphatase deficiency
Genetics	AR	AR	AR	X-linked
Proportionate dwarfism	Yes	Yes	Yes	Yes
Mental Retardation	No	Yes	Yes	Yes
Studies	Keratan sulfate in urine	Dermatan sulfate in the urine	Heparan sulfate in the urine	Dermatan/heparan sulfate in urine
Prognosis	Type A is more severe Type A and B survive into adulthood	Death in first decade of life	Death in second decade of life	Death in second decade of life

Most Common

Mucopolysaccharidoses

- proportionate dwarfism
- increased rate of CTS
- C1-C2 instability
- Hip dysplasia
- abnormal epiphyses
- bullet-shaped phalanges
- genu valgum





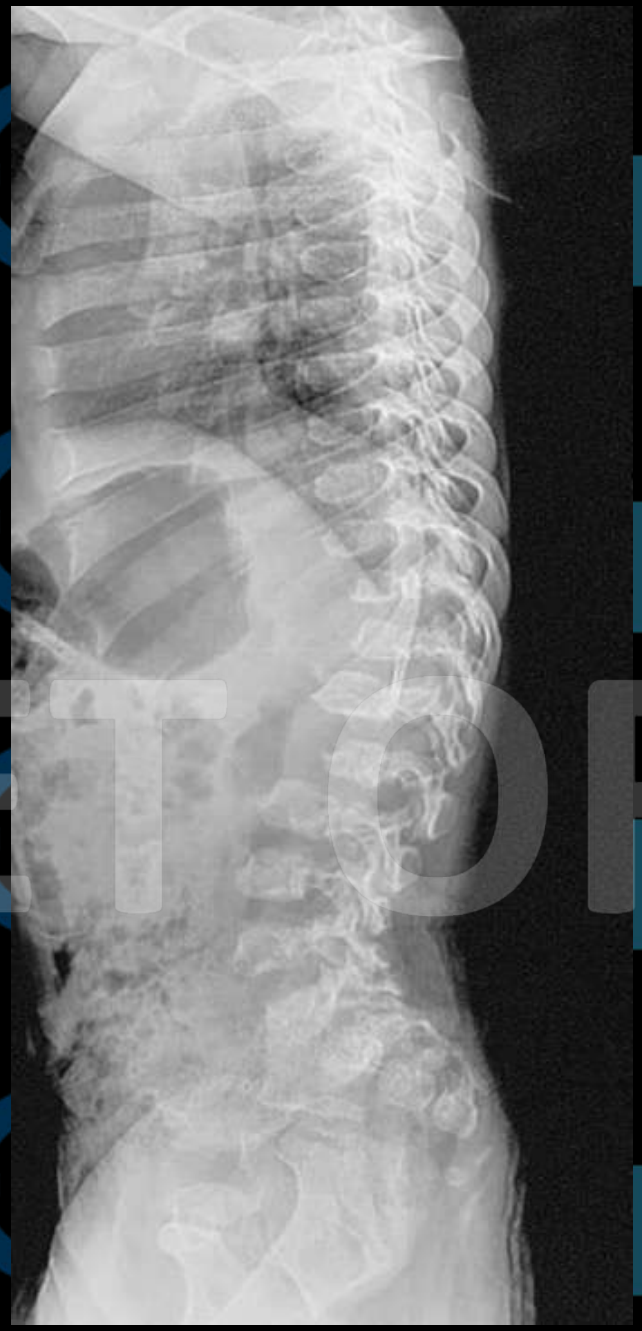
Lysosomal Enzyme which Breaks GAG

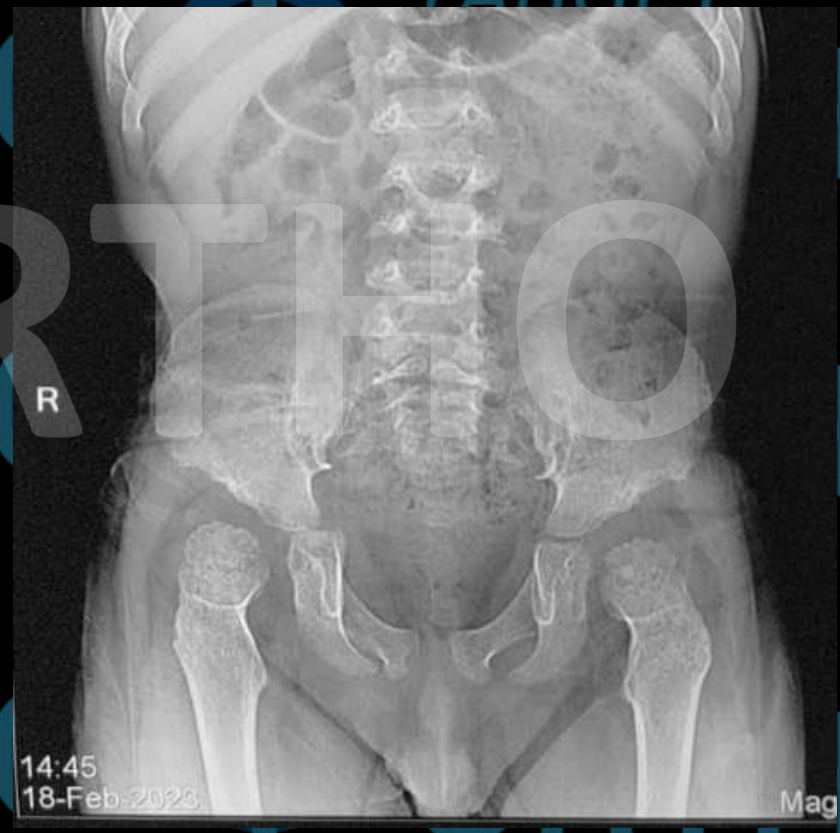
MORQUIO SYNDROME

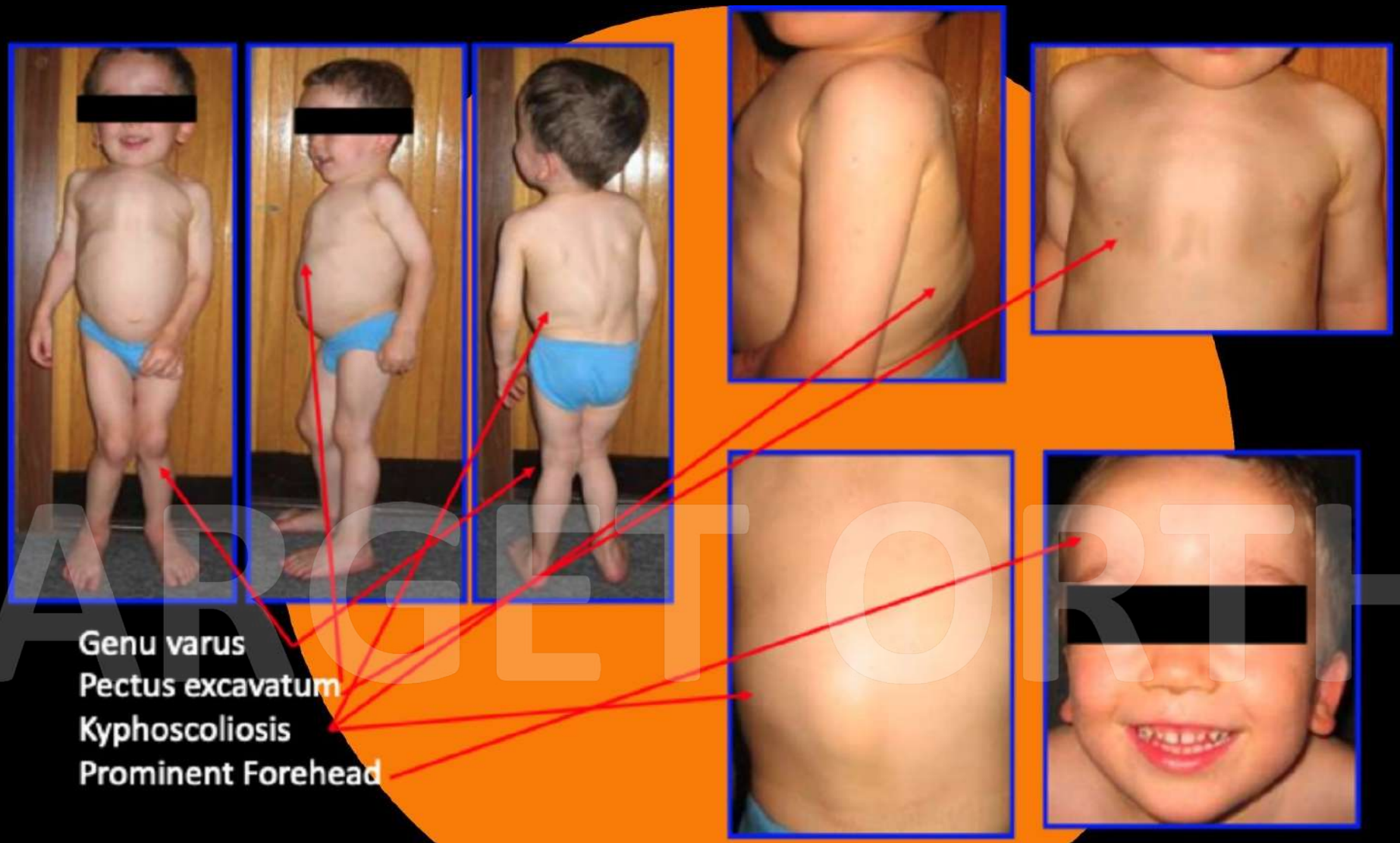
- Accumulation of keratan sulfate
- interferes with the cartilage at the growth plate

Pathophysiology:
Type A (galactosamine-6-sulfate-sulphatase deficiency)
Type B (beta-galactosidase deficiency)

Genetics
AR;
Type A is more severe







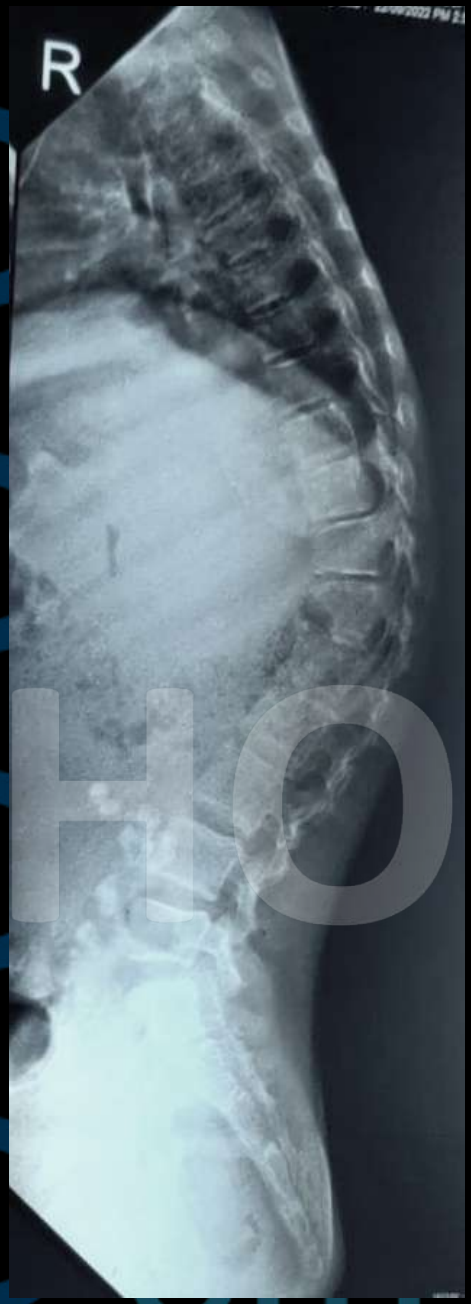
https://emedicine.medscape.com

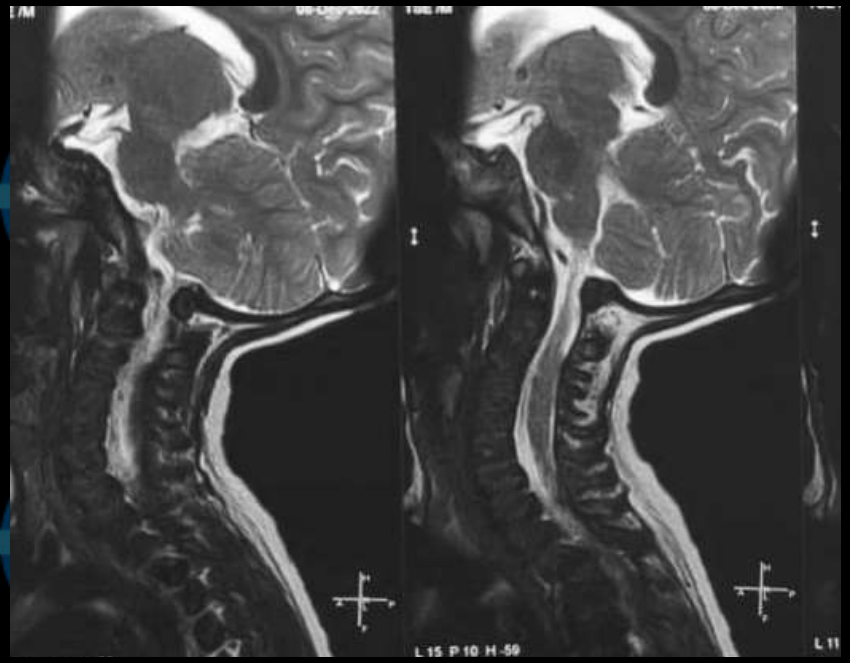
MORQUIO'S

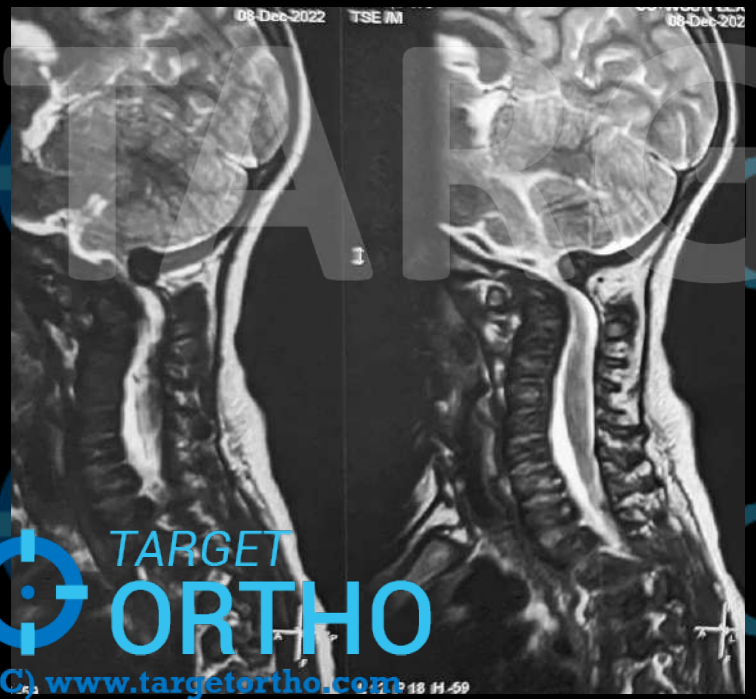
Gaucher Disease

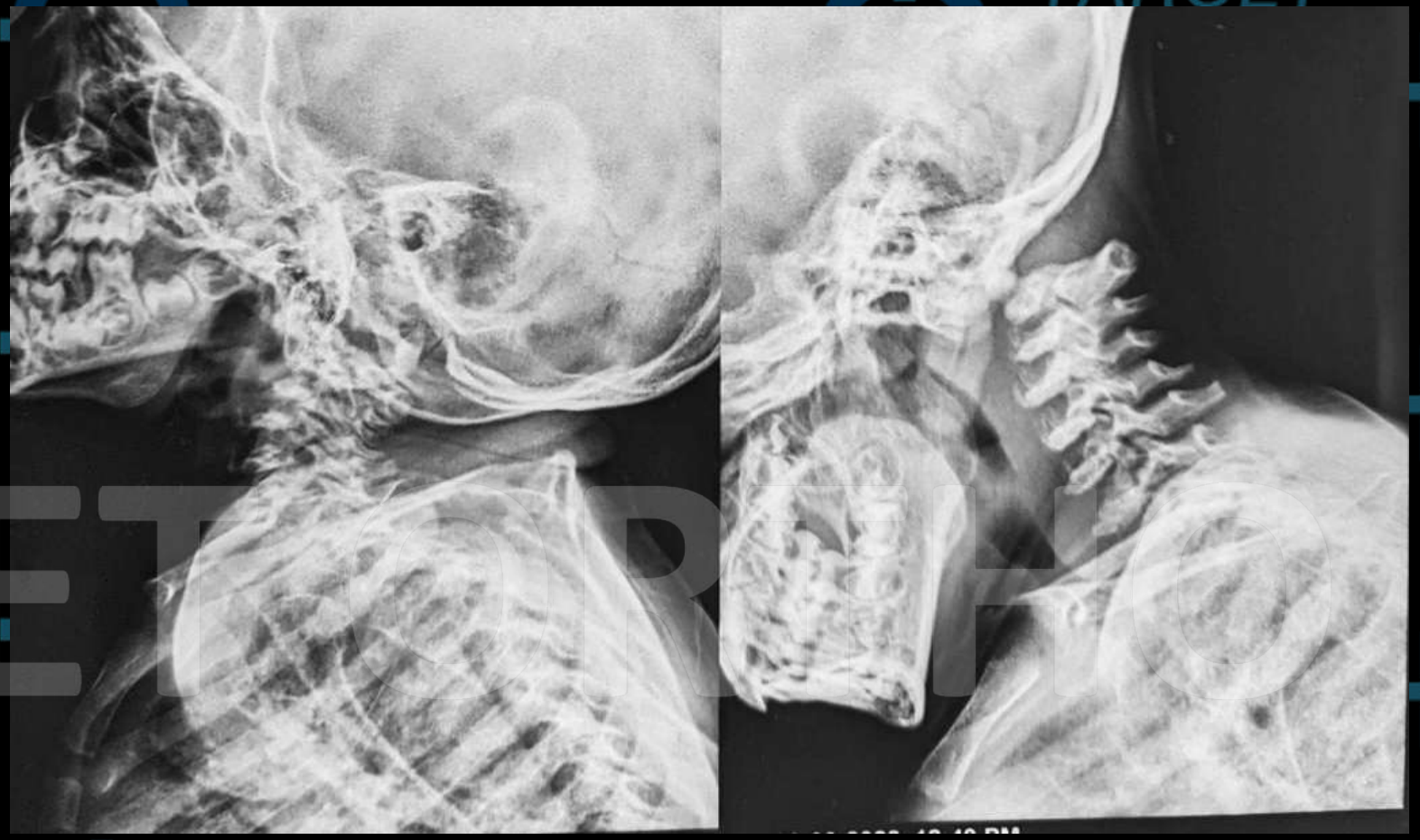
- Kyphosis with Liver enlargement
- Accumulation of glucocerebroside in the reticuloendothelial system

- Osteoporosis >> Fracture and wedging of vertebrae
- Hepato/ Splenomegaly
- Progressive Kyphosis





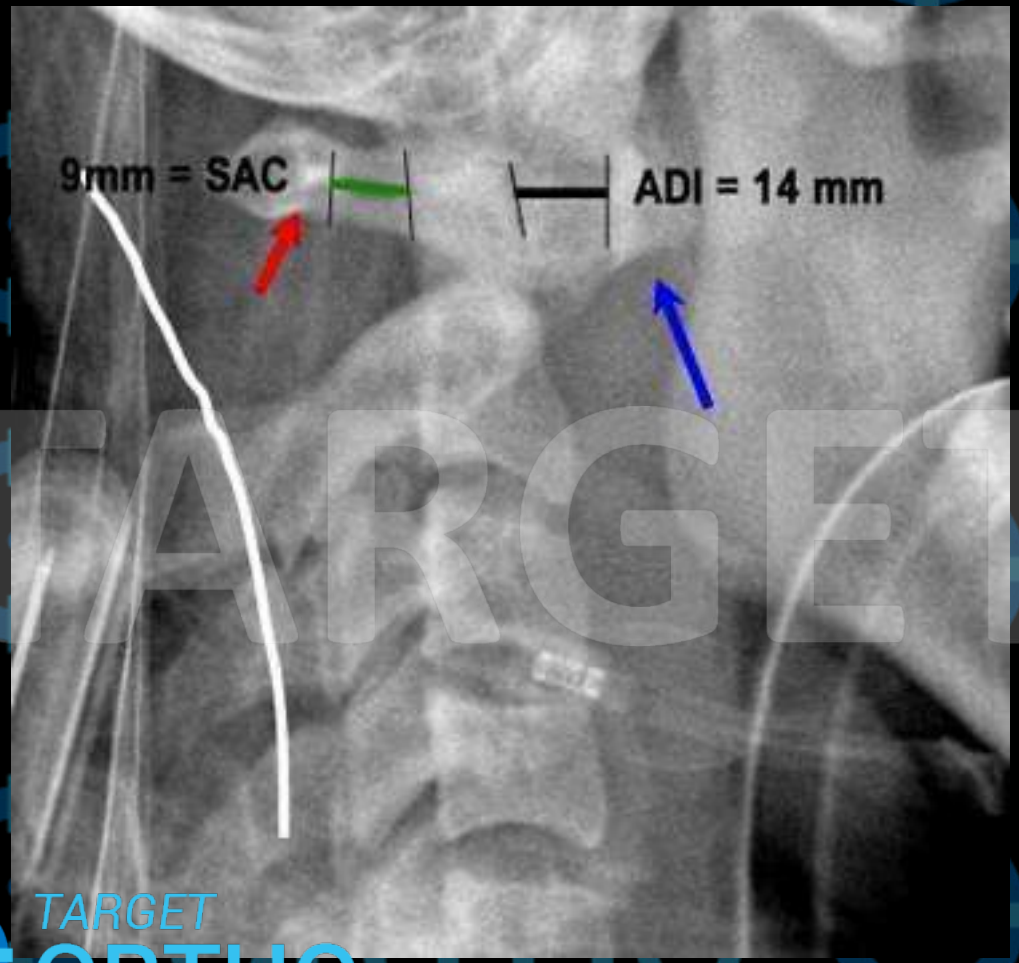


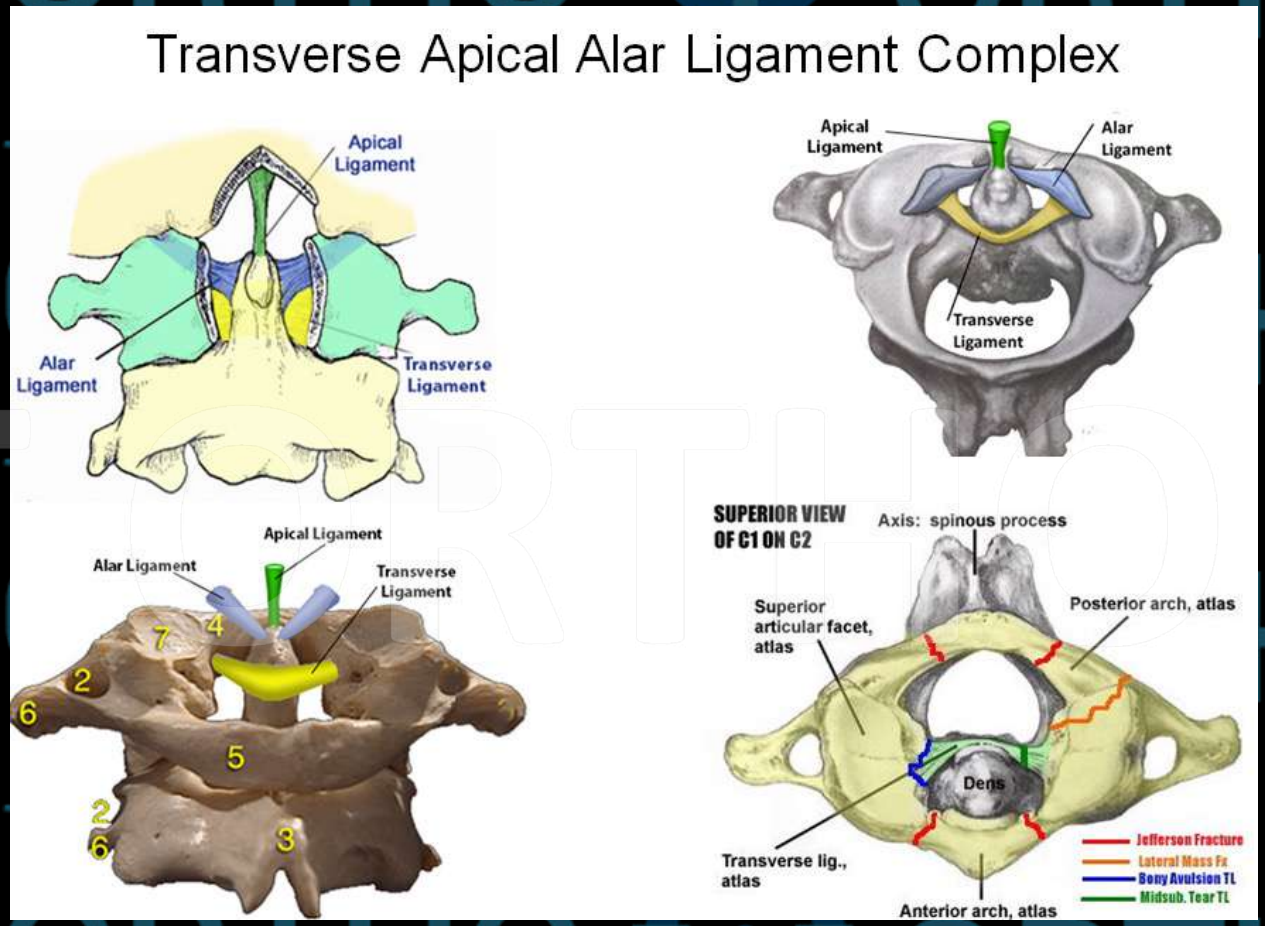


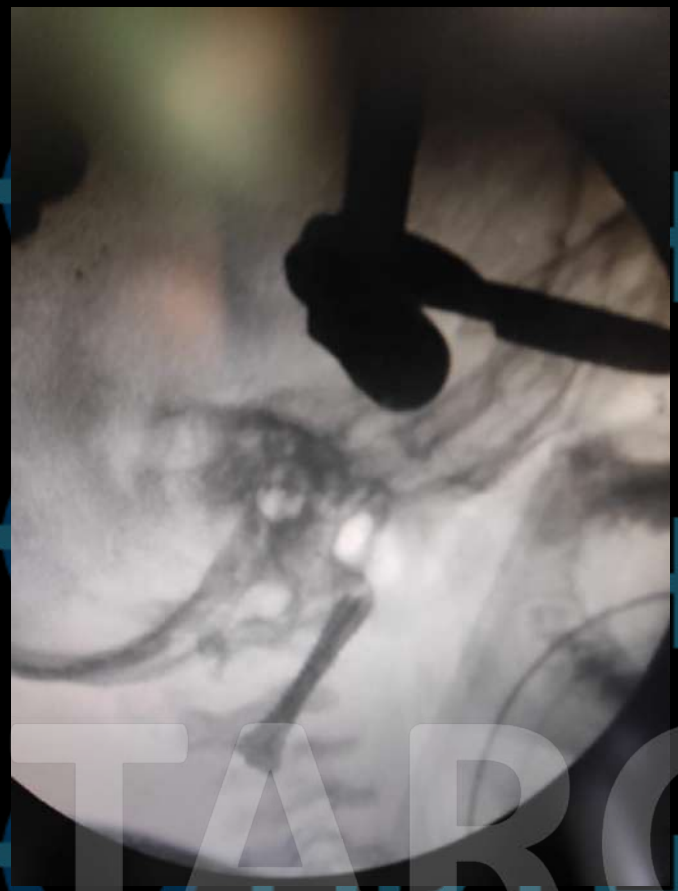
Neurology Important

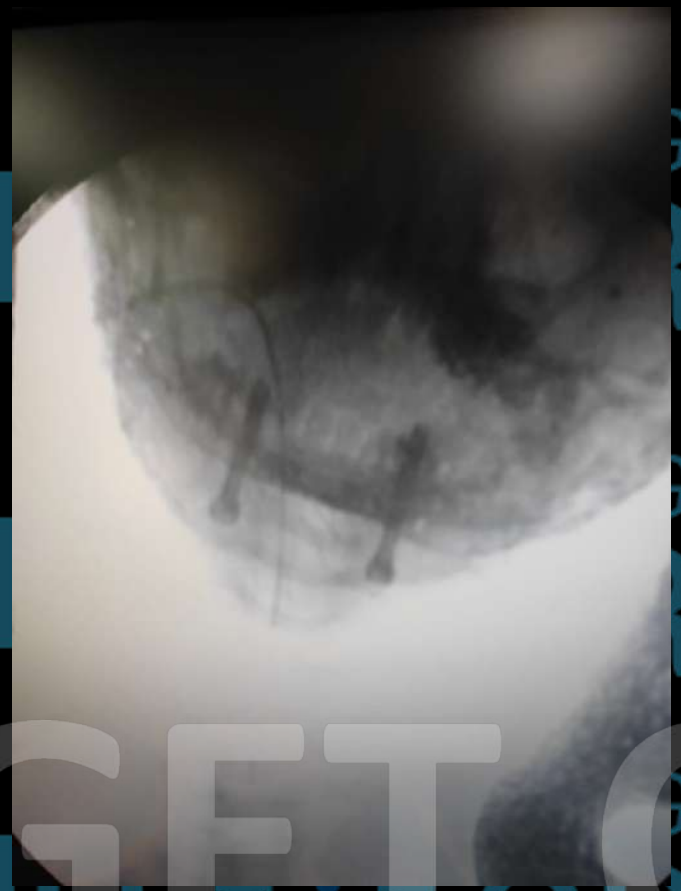
- Hyperreflexia (patellar tendon reflex)
- muscles weakness
- broad based gait
- decreased hand dexterity
- loss of motor milestones
- bladder problems

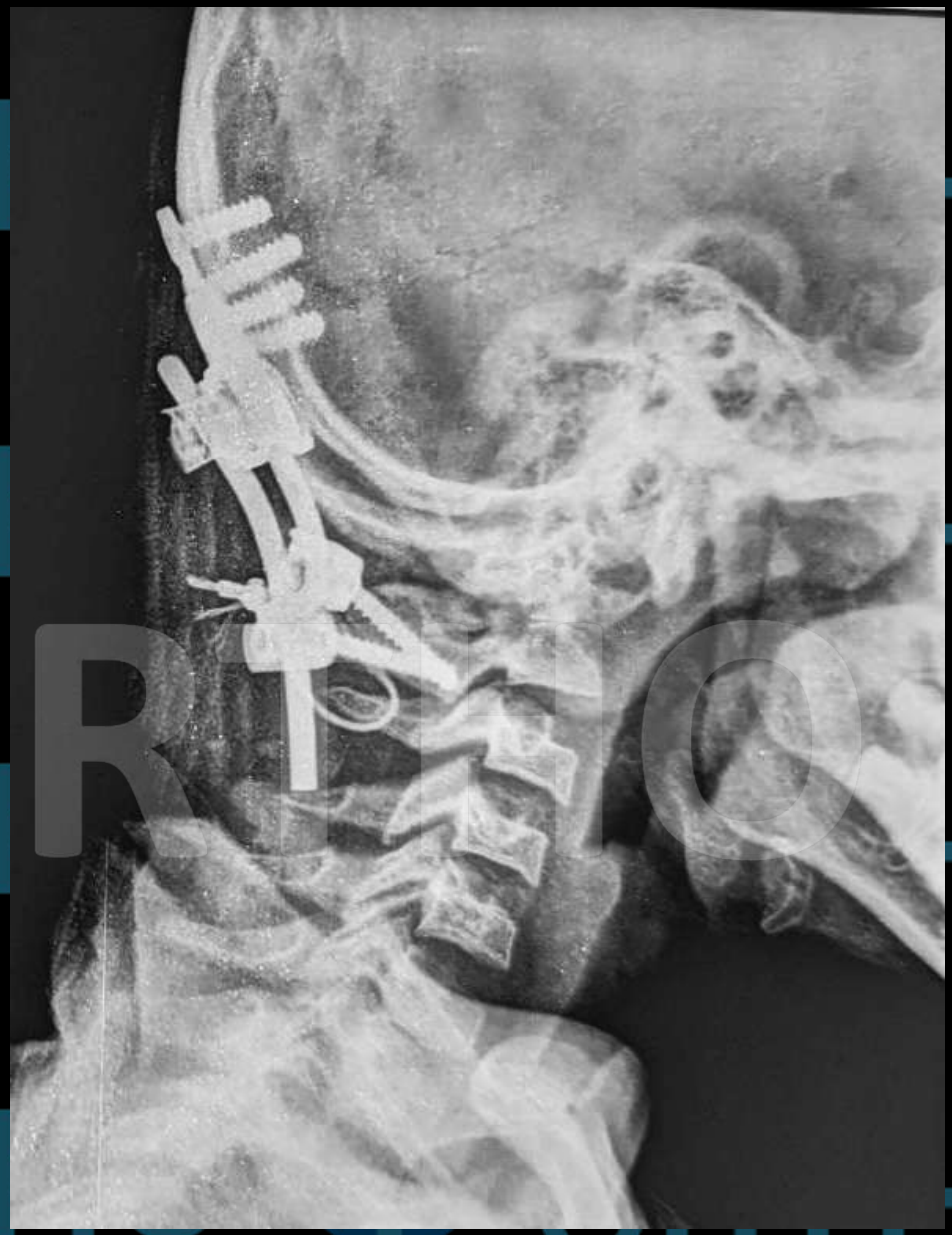
Atlantoaxial instability

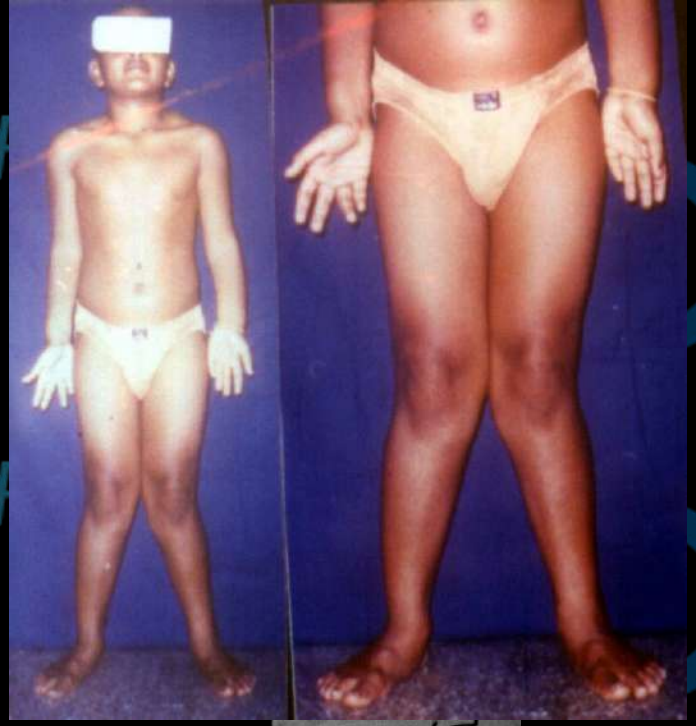




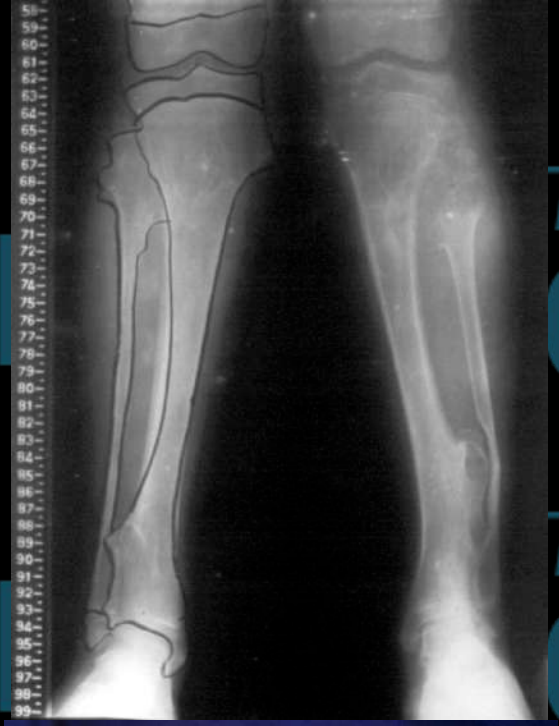


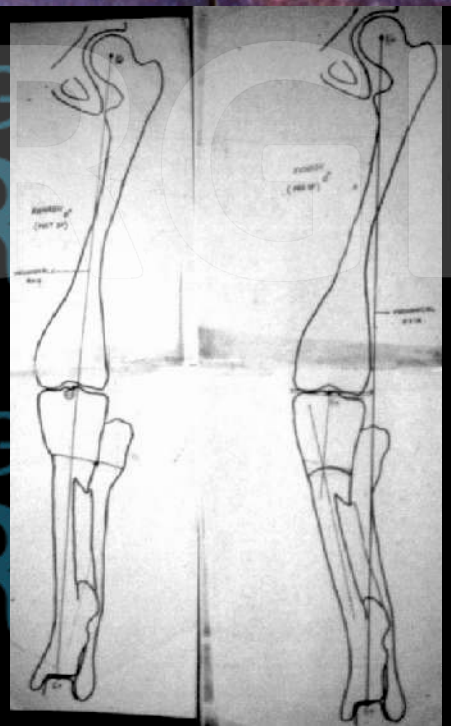


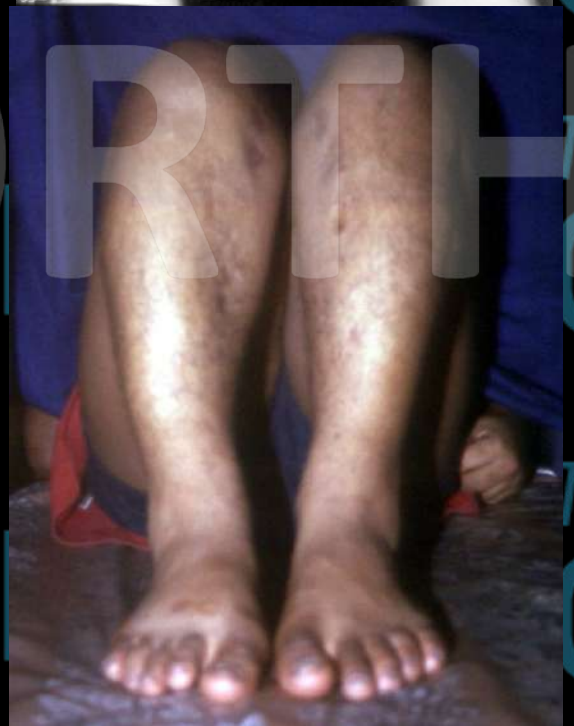


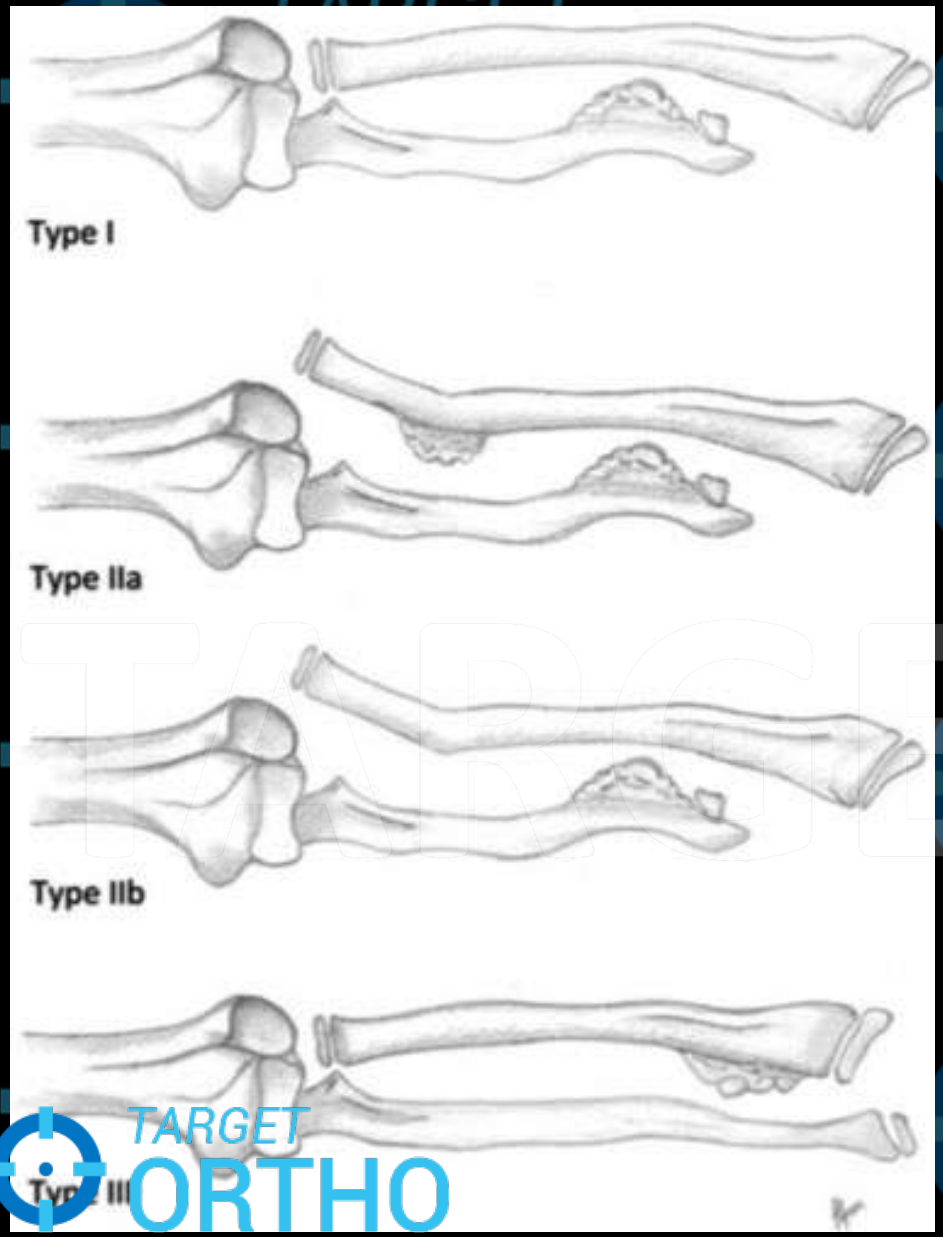


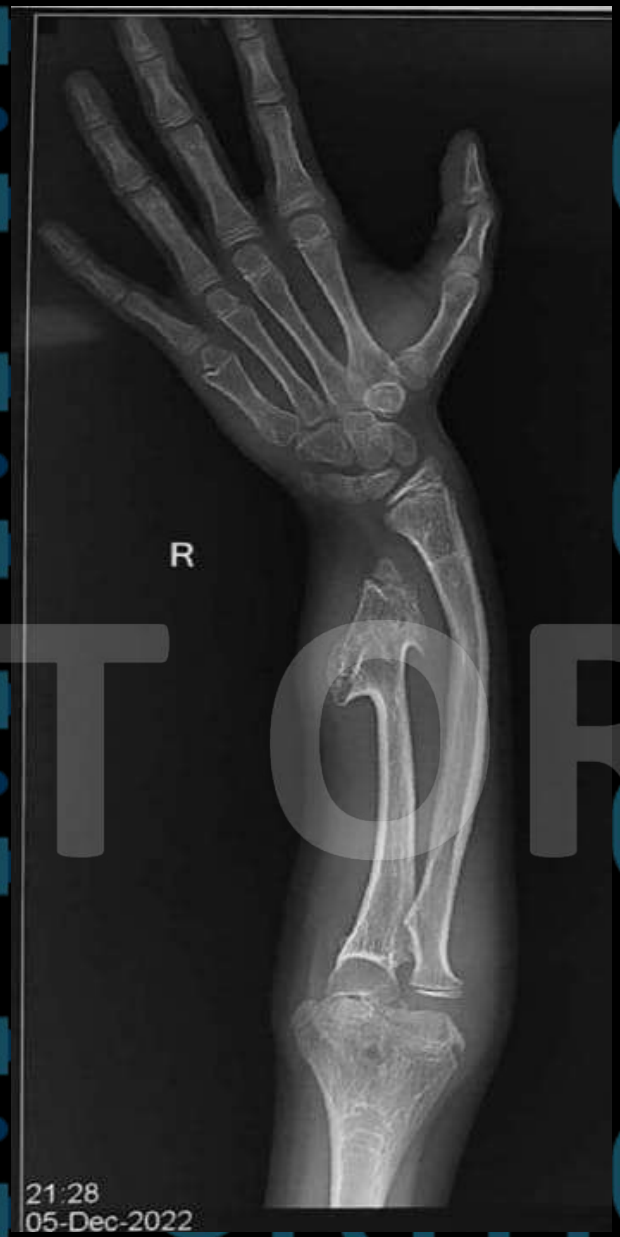
Hereditary Multiple Exostosis



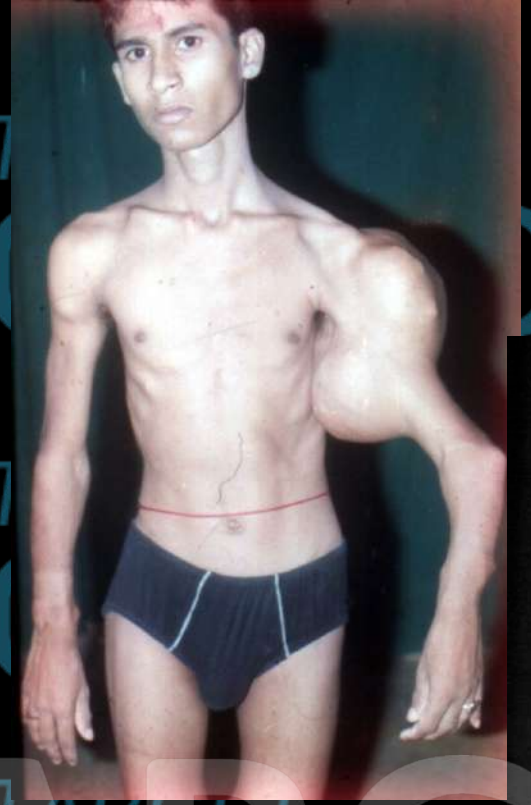


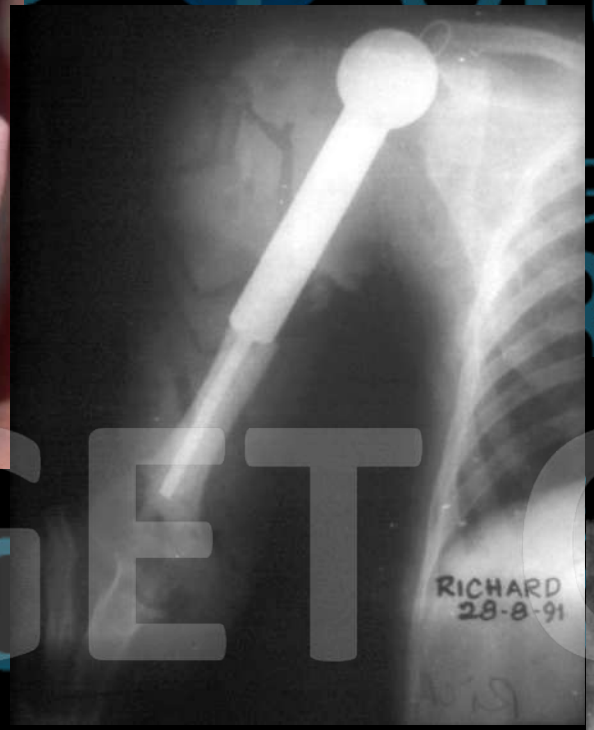


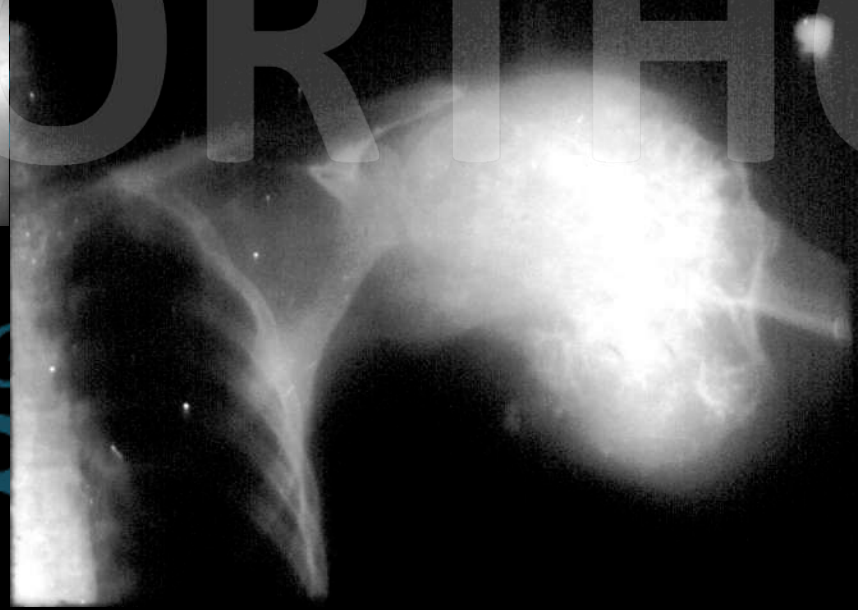


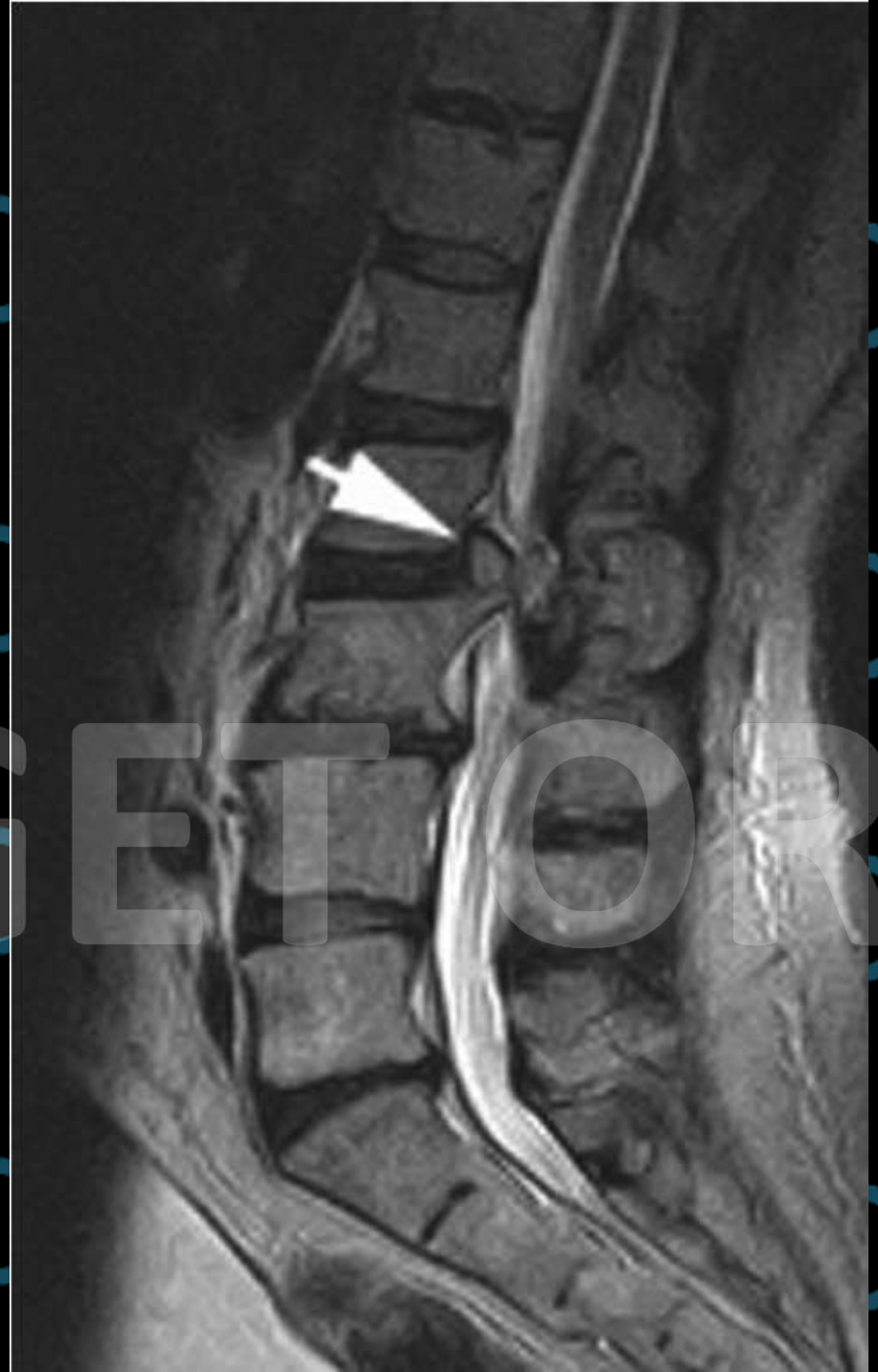


Multiple Exostosis

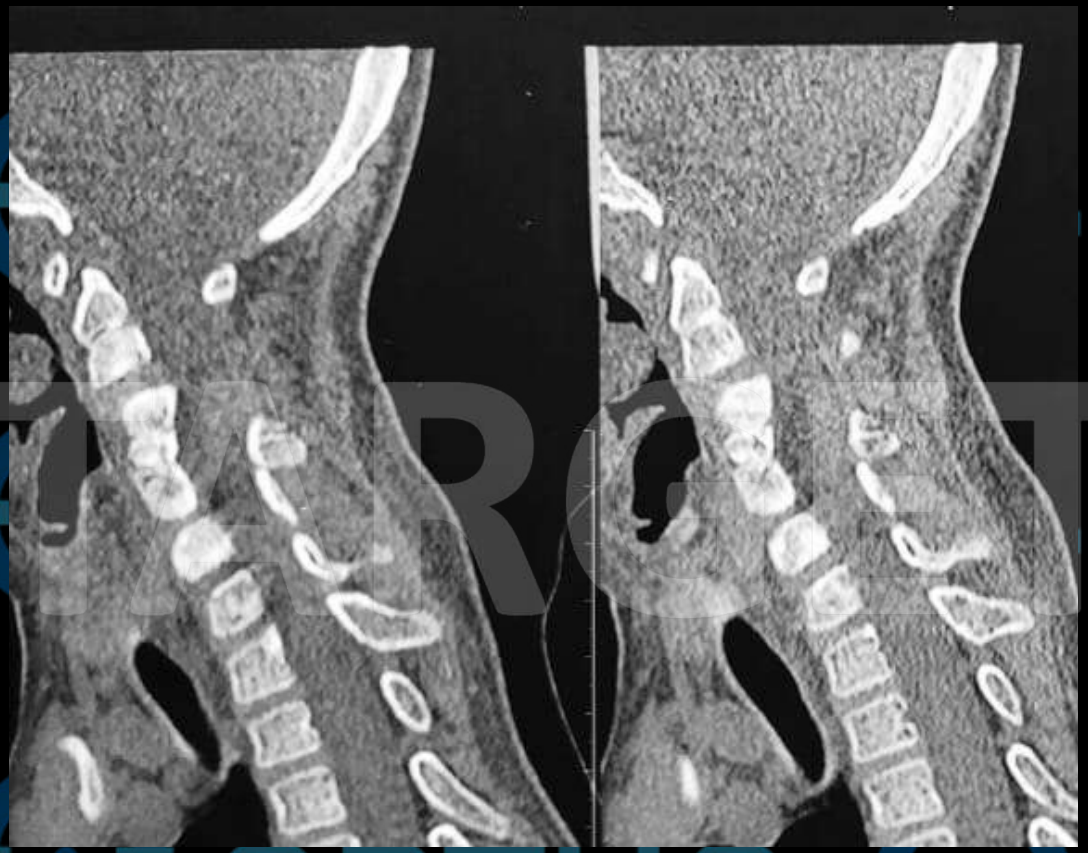








Klippel Feil



DIAGNOSTIC EVALUATION OF SKELETAL DYSPLASIA

1. RECOGNISE SHORT STATURE
2. DETERMINE SITE OF DISPROPORTION
3. DETERMINE SEGMENT OF LIMB
4. IDENTIFY DYSMORPHISM AND DEFORMITY

DIAGNOSTIC EVALUATION OF SKELETAL DYSPLASIA

5. CATEGORISE THE RADIOLOGICAL INVOLVEMENT

6. LAB EVALUATION, IF NECESSARY

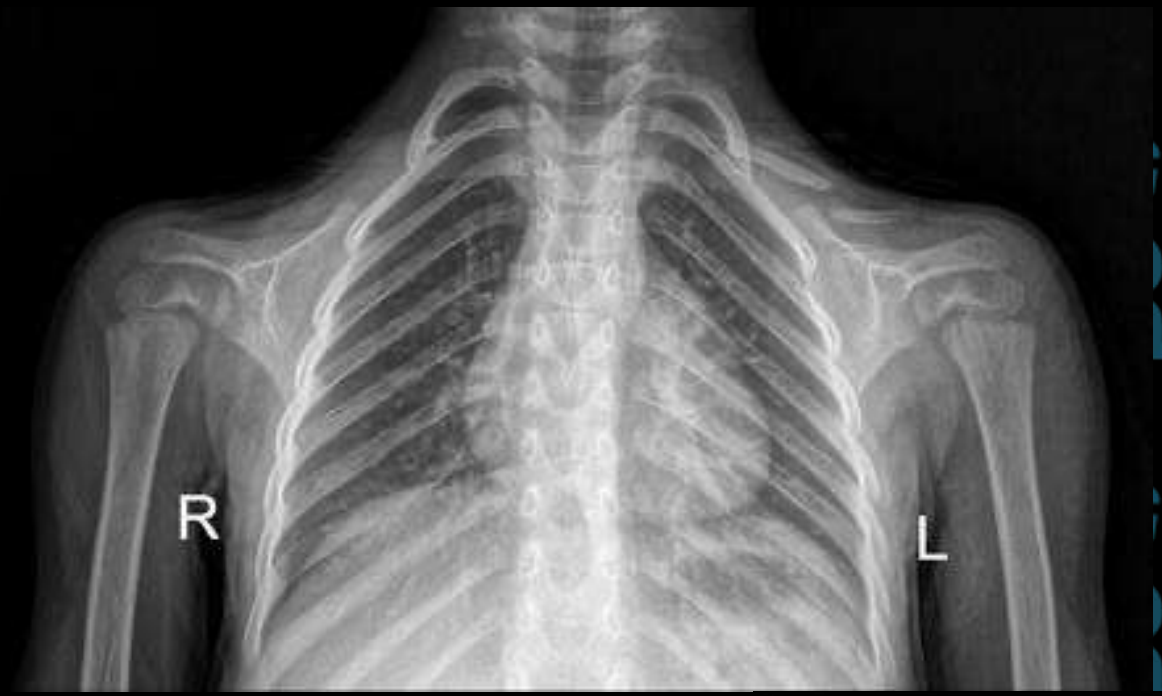
7. FAMILY PEDIGREE

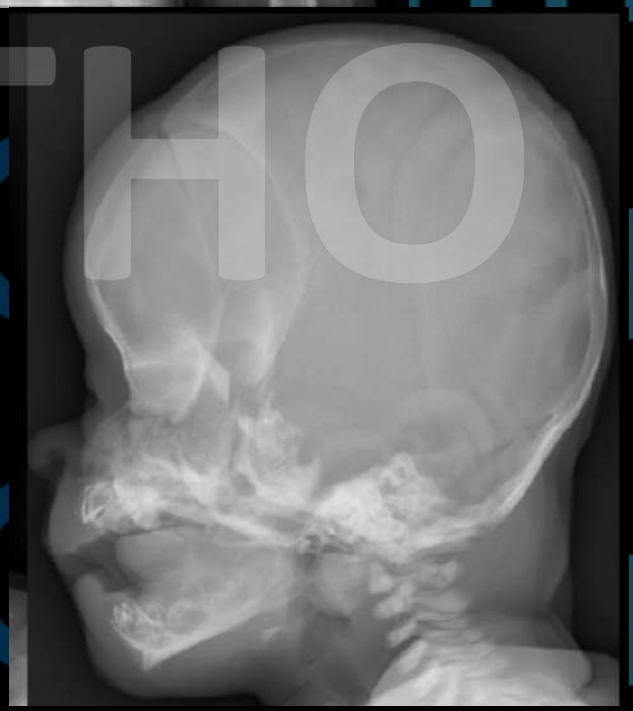
8. PERIODIC EXAMINATION IF DIAGNOSIS IS UNCLEAR

9. CONSULTATION WITH

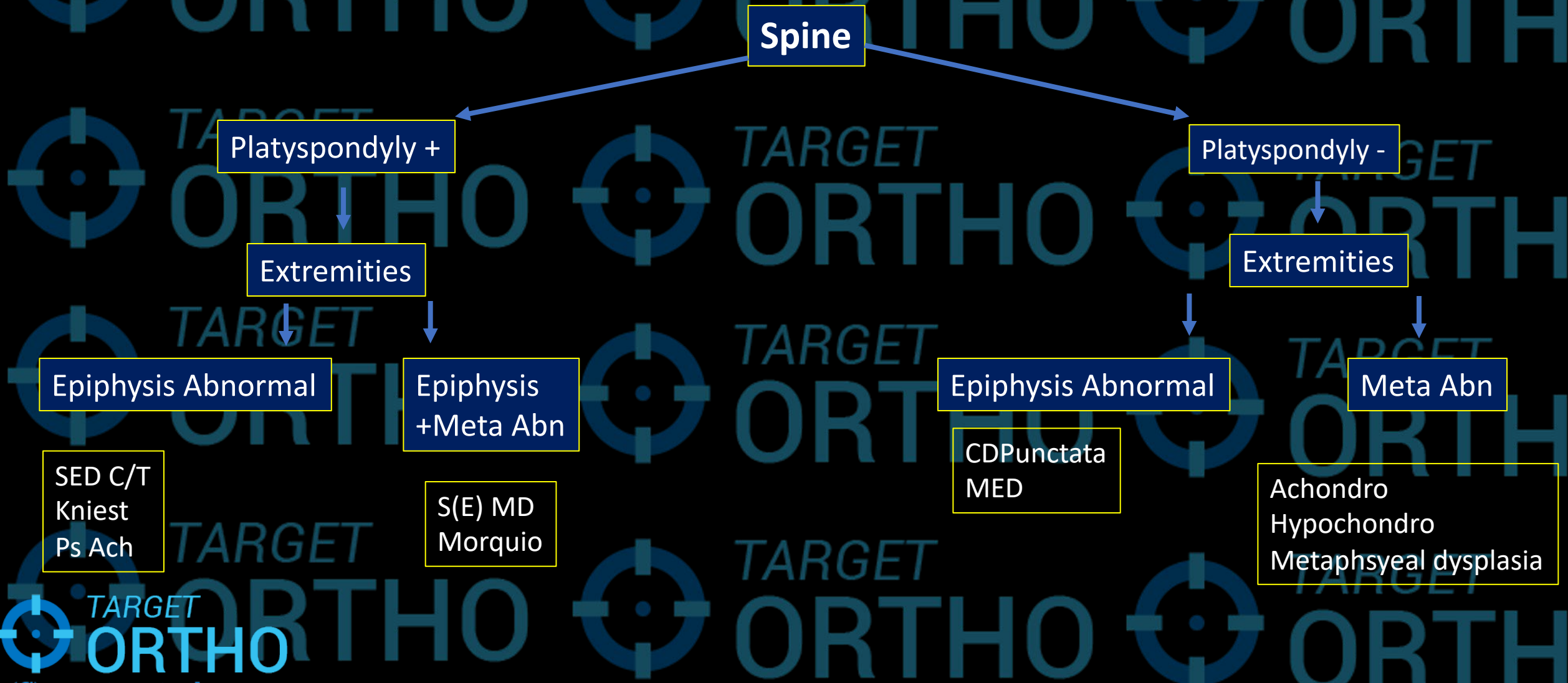
Skeletal Survey

- Skull Lateral
- Chest PA
- LS Spine Lateral
- Pelvis with Hips
- Long bones LL AP
- Hands AP





Skeletal Survey



Skeletal Survey

The Big 10

- Achondroplasia
- MPS
- SED
- Metaphyseal Dysplasia
- Fibrous dysplasia
- Osteopetrosis
- Craniosynostosis and variants
- Klippel Feil
- Diaphysial aclasia (HME)
- Osteogenesis imperfecta

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