

56th Annual Meeting &
42nd Post Graduate Course

June 06 - 10 2022
Symposium Sanofi



ESPR
European Society of
Paediatric Radiology

Marseille

METABOLIC DISEASES AND BONE DYSPLASIAS : RADIOLOGIST'S EYE

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ESPR 2022 June 9th, Marseille

LINK OF INTERESTS

SANOFI : speaker and scientific committee honorary



ORIENTATION OF THE
DIAGNOSIS BY THE
RADIOLOGIST ?

Whistleblower....?

- At the crossroads of specialities:
pediatrician, endocrinologist, rheumatologist,
orthopedist, cardiologist,
- Can identify metabolic storage
diseases
- Patients followed by different
specialists for various symptoms
- According to clinical signs gradually
developed : imaging x-rays
requested



EXAMPLE OF RARE METABOLIC STORAGE DISEASES : MPS

1/25 000-1/30 000 live births

- Specific enzyme deficiency => accumulation of glycosaminoglycans (GAGs)=> progressive multi tissue damage
- Screening : difficult.....
- Biological diagnostic orientation : easy !! measure the urinary GAGs
- Dg confirmed => leucocytes enzymes



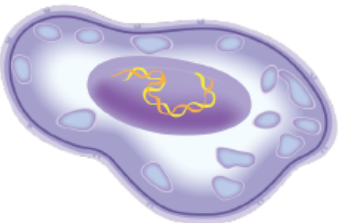
ACCUMULATION OF GAG

- **CHONDROBLASTS:**
SECONDARY OSSIFICATION
DEFECT= multiple dysostosis

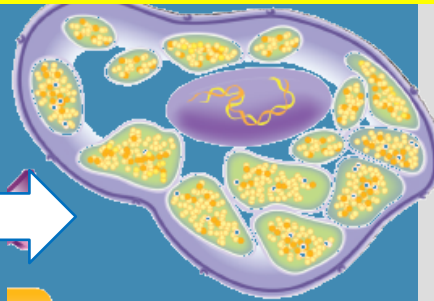
- **FIBROBLASTS : CONNECTIVE**
TISSUE INFILTRATION (ligaments,
tendons and capsules)=> notch

Normal cell

Cellule normale



Cell loaded with GAG





MPS I

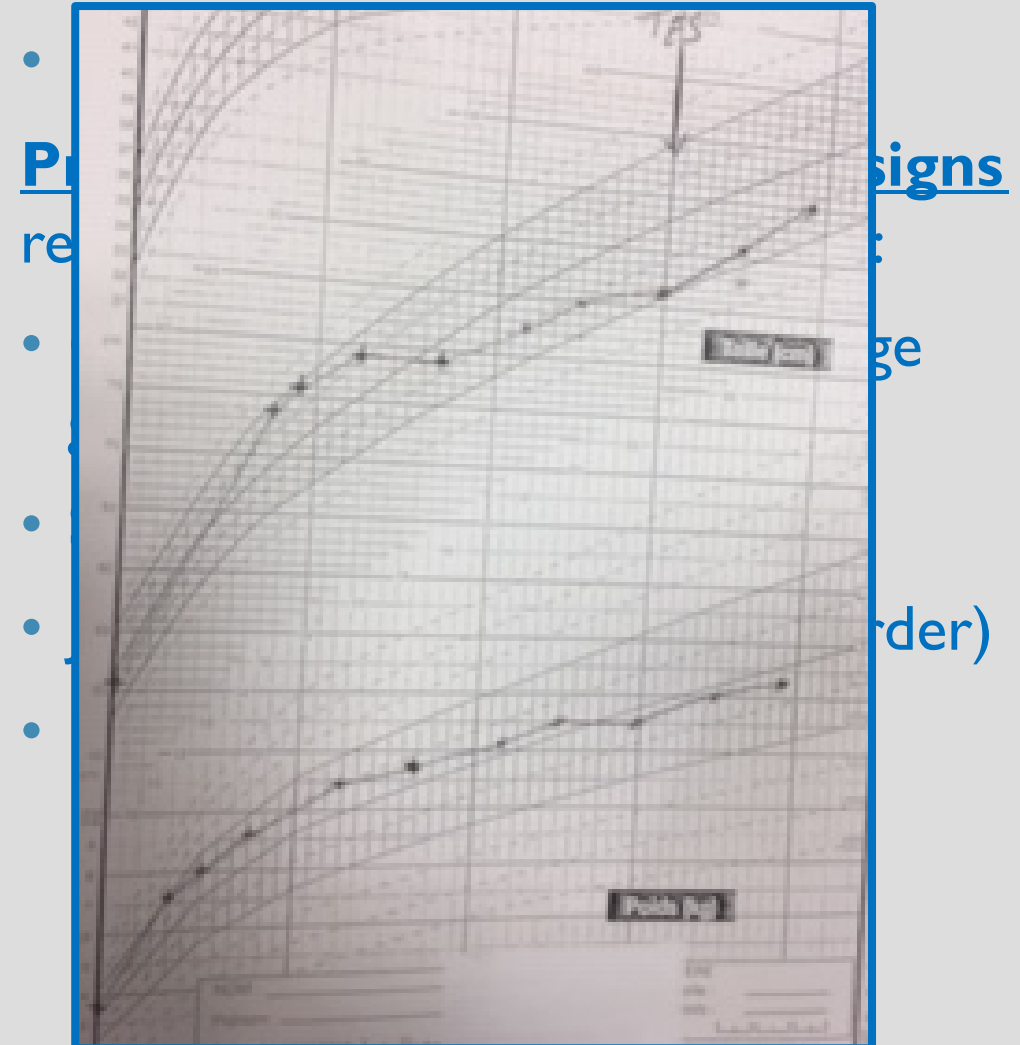
ENZYME= ALPHA-L-IDURONIDASE

3 phenotypes: Hurler
(severe), Hurler-Scheie
(intermediate form), Scheie
(moderate)

7-8 new cases* MPS I / year in France

23 MPS/year

**D'après les cahiers d'Orphanet : Prévalence des maladies rares Juin 2018 et 738000 naissances en France INSEE*



- **P**revalence
- **re**gion
- **signs**
- **age**
- **order**

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SO, JUST THINK ABOUT IT

....

Awareness
of the Radiologist

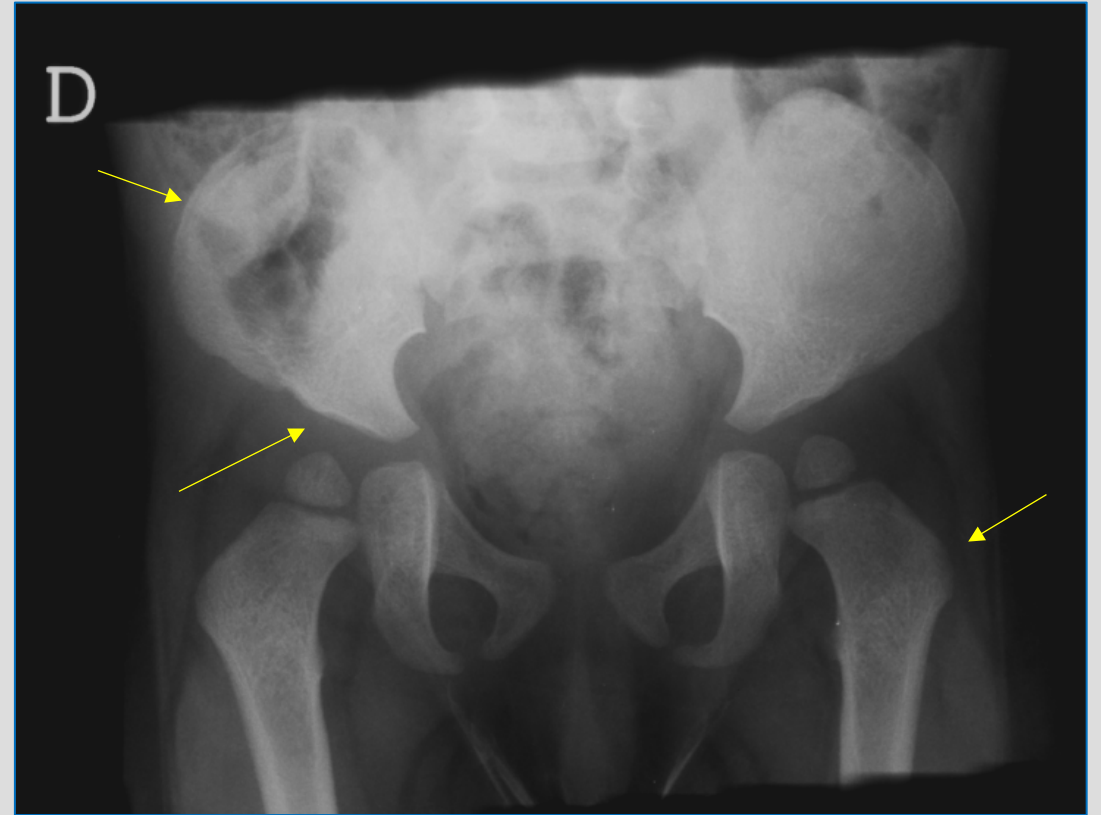
- Identify multiple dysostosis
- young children



- Pelvic x-ray
- Full spine
- Wrist and hand x-ray (bone age study)



FRONT VIEW PELVIC X-RAY

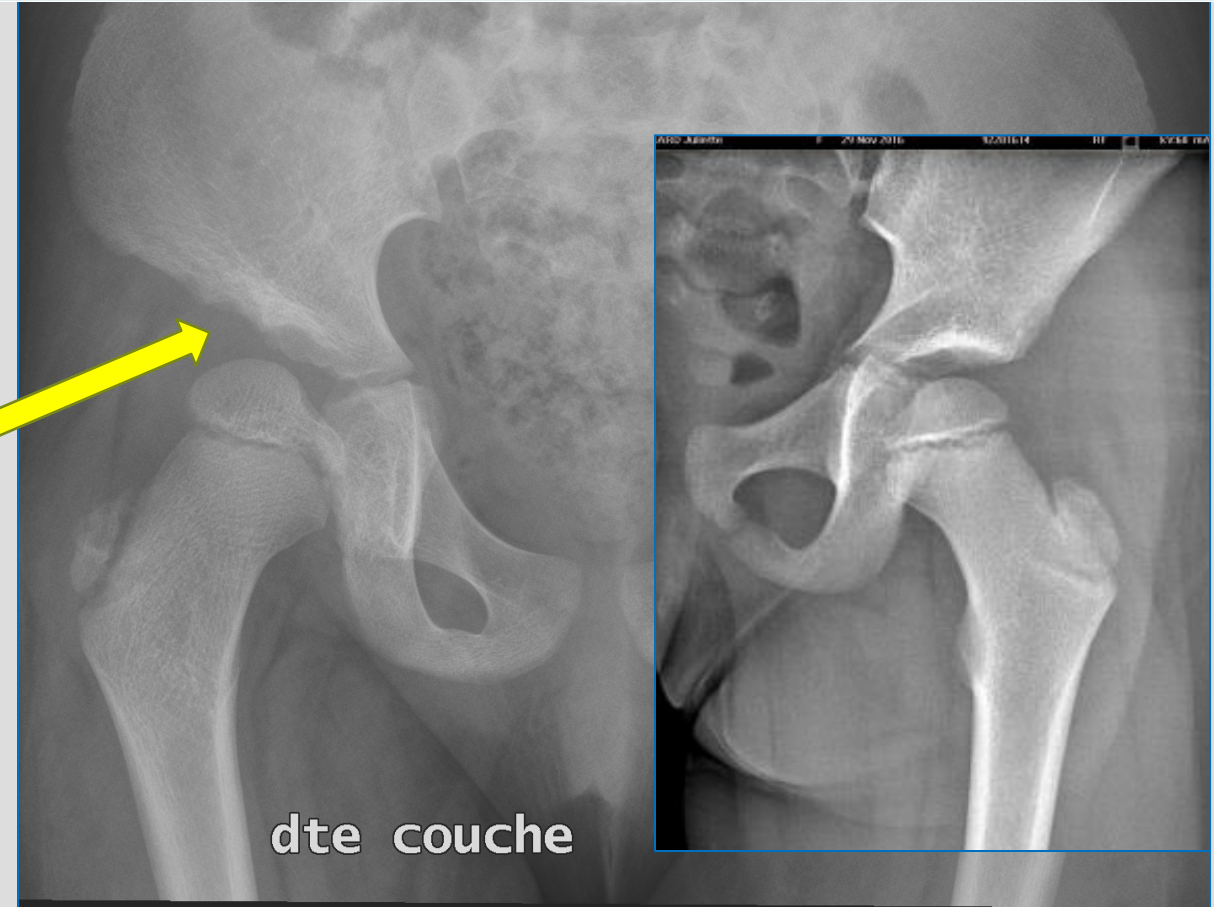
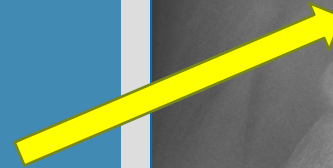


Girl, 13 mois, Hurler (Courtesy Pr Panuel)

Rounded iliac wings
Acetabular dysplasia
Coxa valga



TYPICAL IMAGING OF
DISEASE



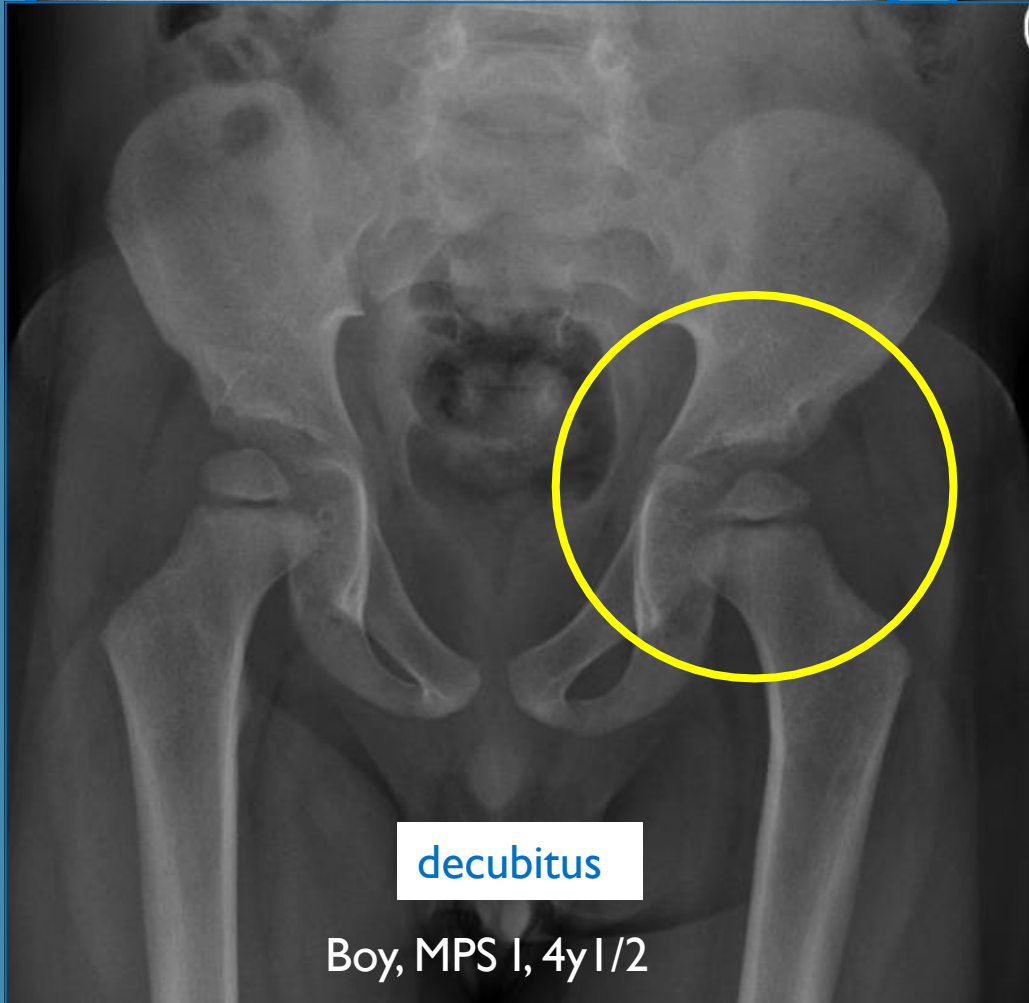
Girl, 5 years, Hurler (courtesy Pr Panuel)

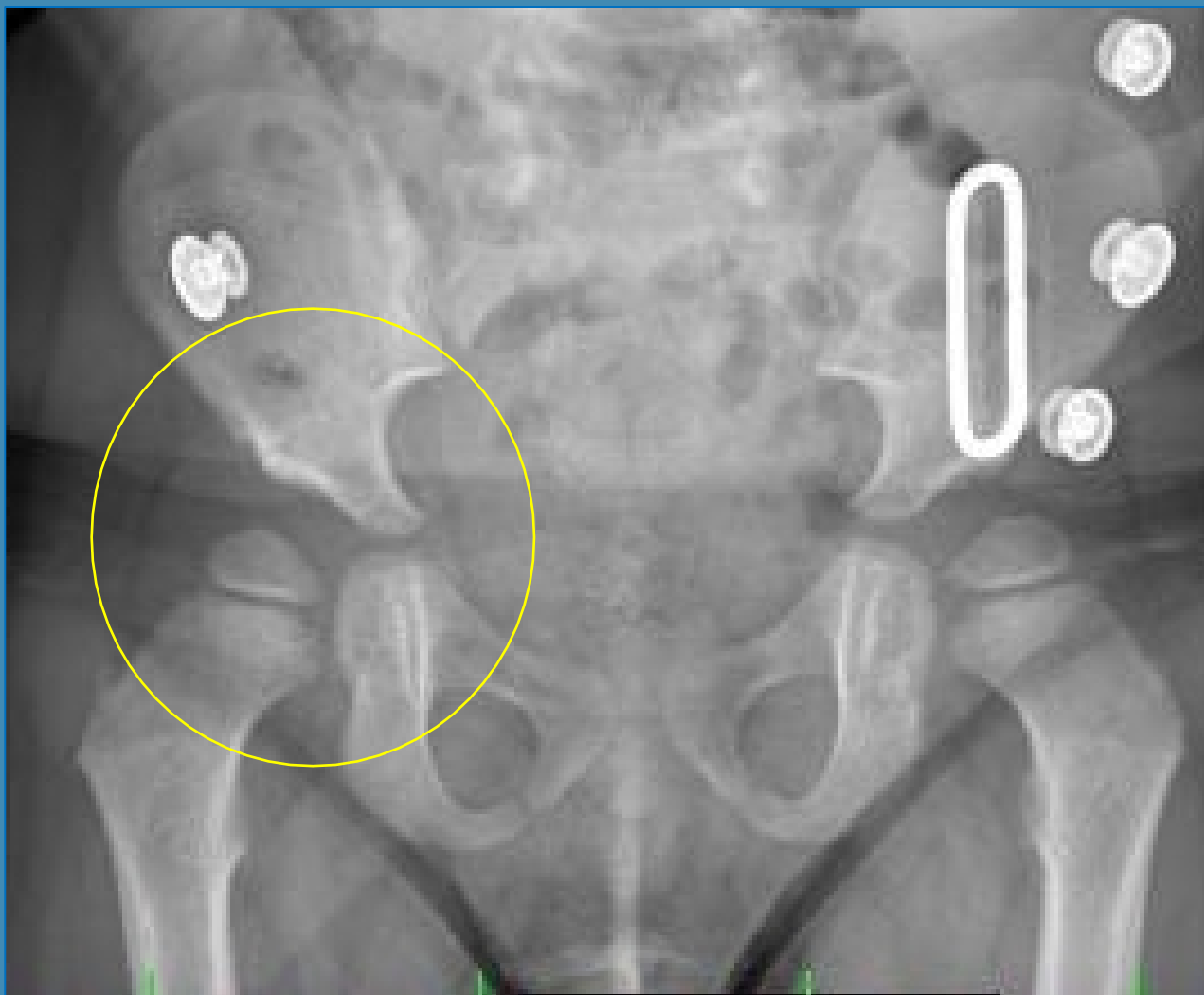
Notch

Underdeveloped proximal femoral epiphysis

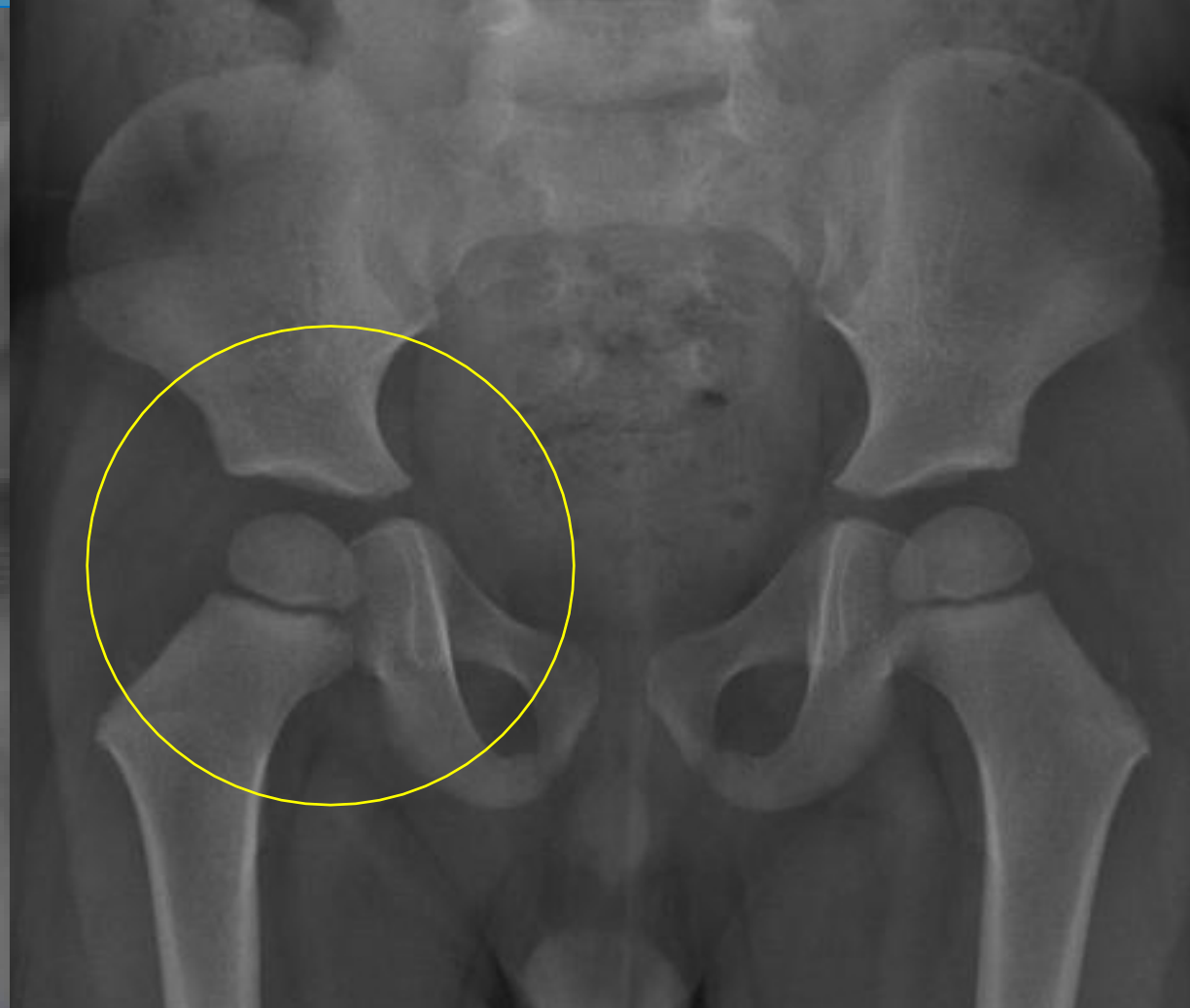


Girl, MPS I, 2y





Boy, 3 ans, MPS II



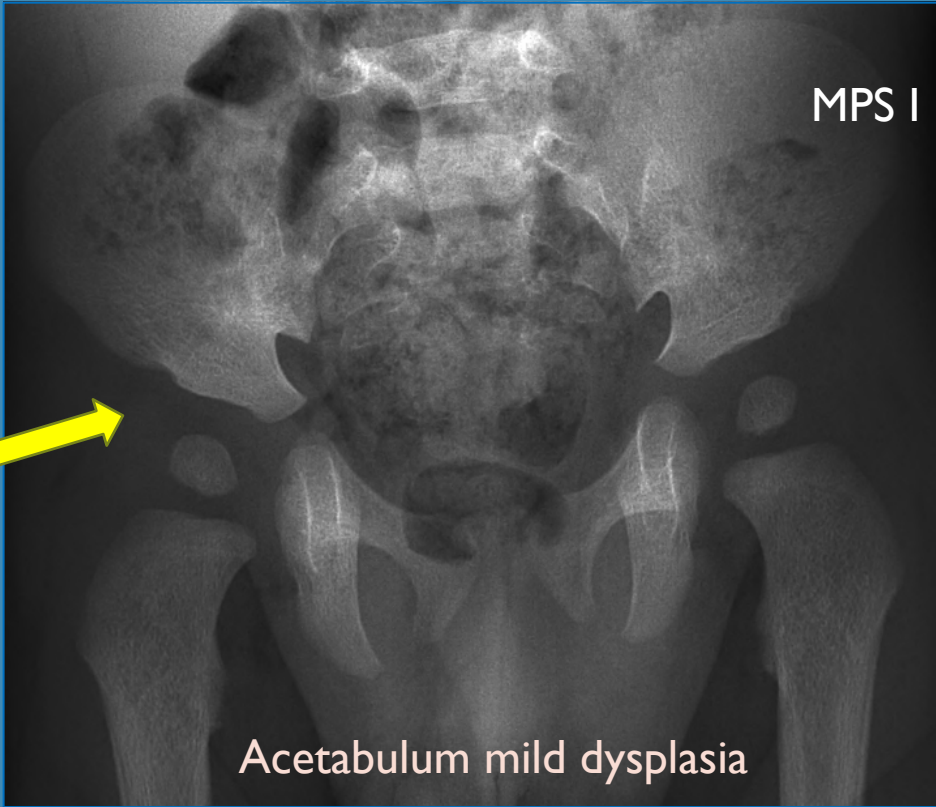
Normal

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MPS I Scheie
Upper acetabulum notch

PELVIC X-RAY



MPS I Scheie



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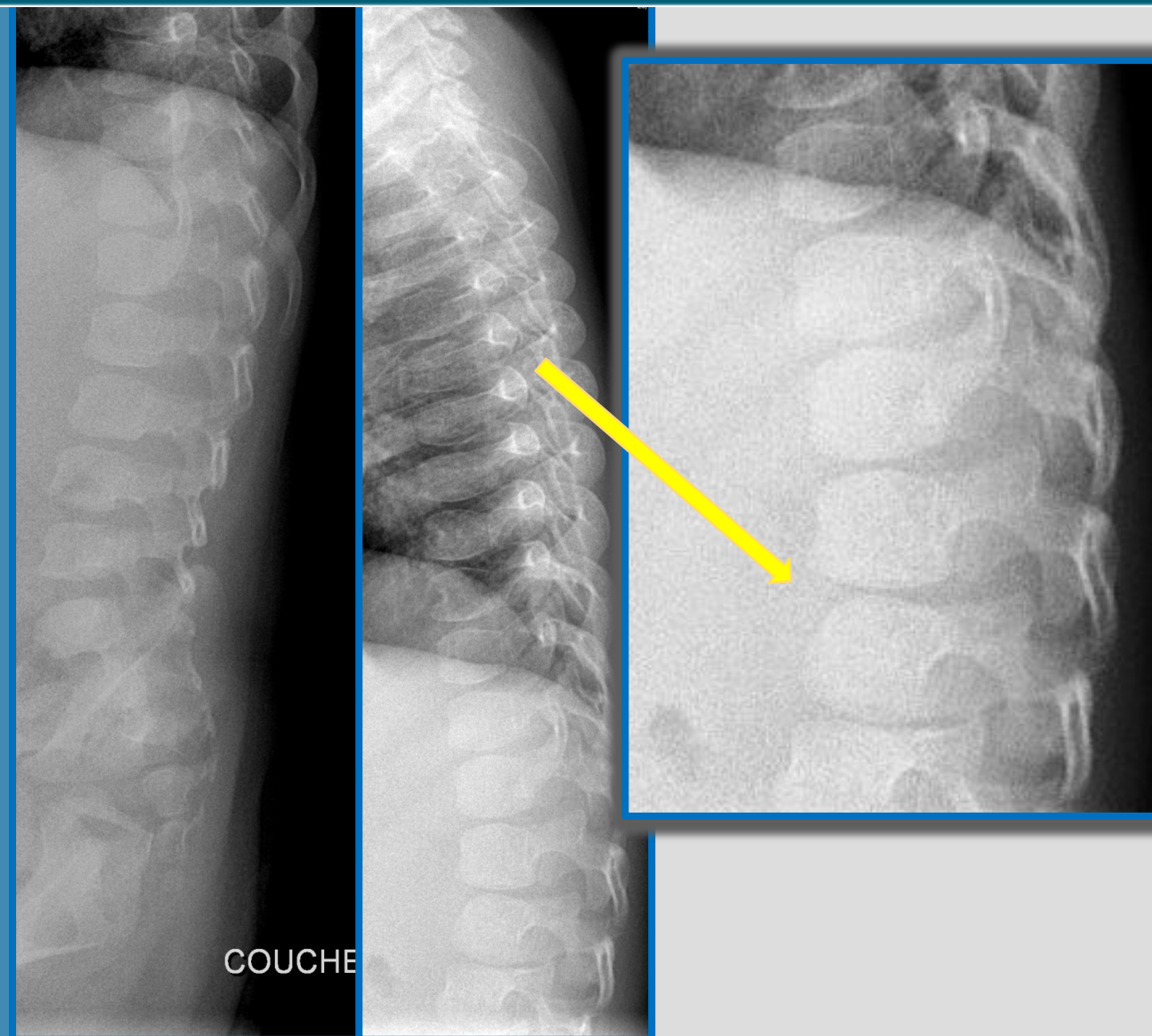
Boy, MPS I, 7y.
Flessum, joint stiffness





PROFILE VIEW OF SPINE

Girl, MPS I , 2y1/2
Rostrum L3
Mild ovoid vertebrae





PROFIL VIEW OF SPINE
UNUSUAL KYPHOSIS

Boy, MPS II, 2y

Rostrum

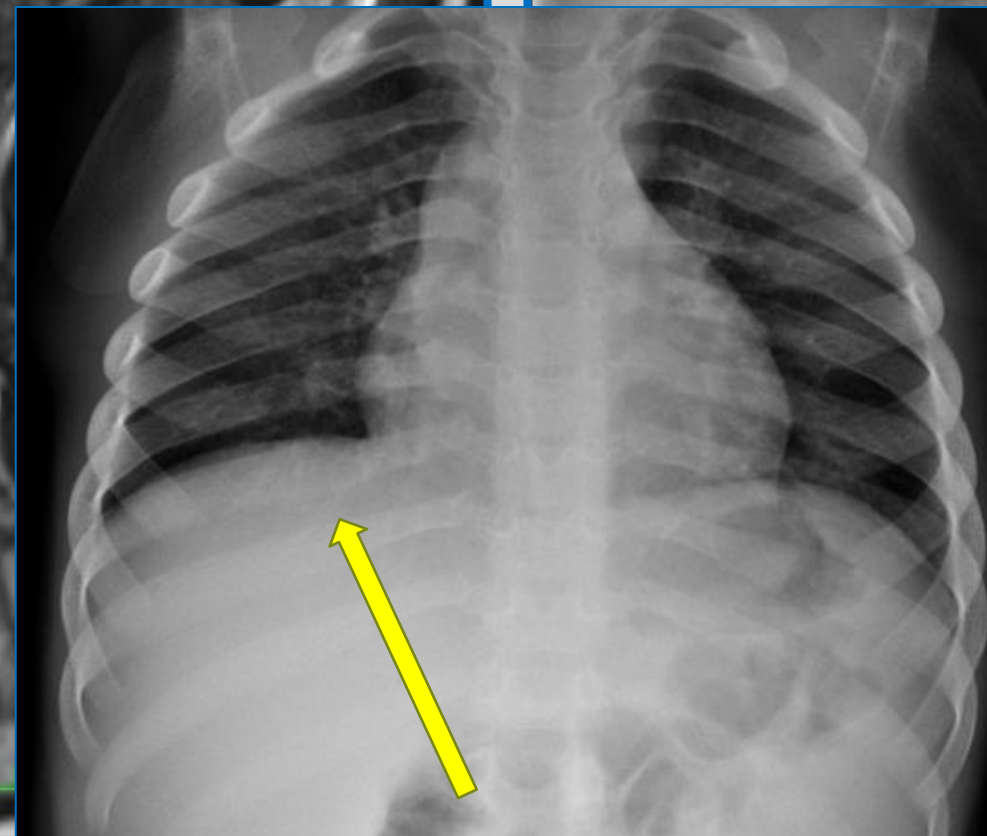
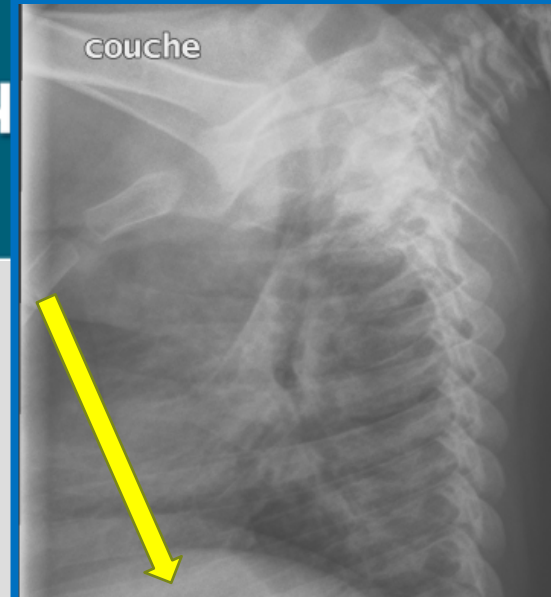
Wide ribs

MRI, T2



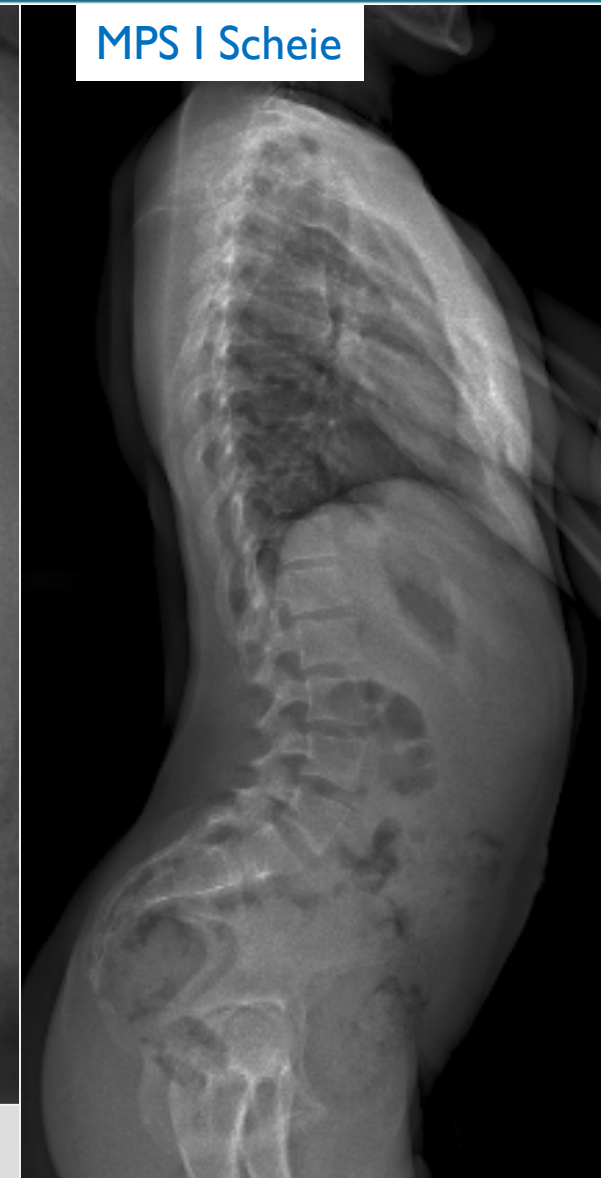
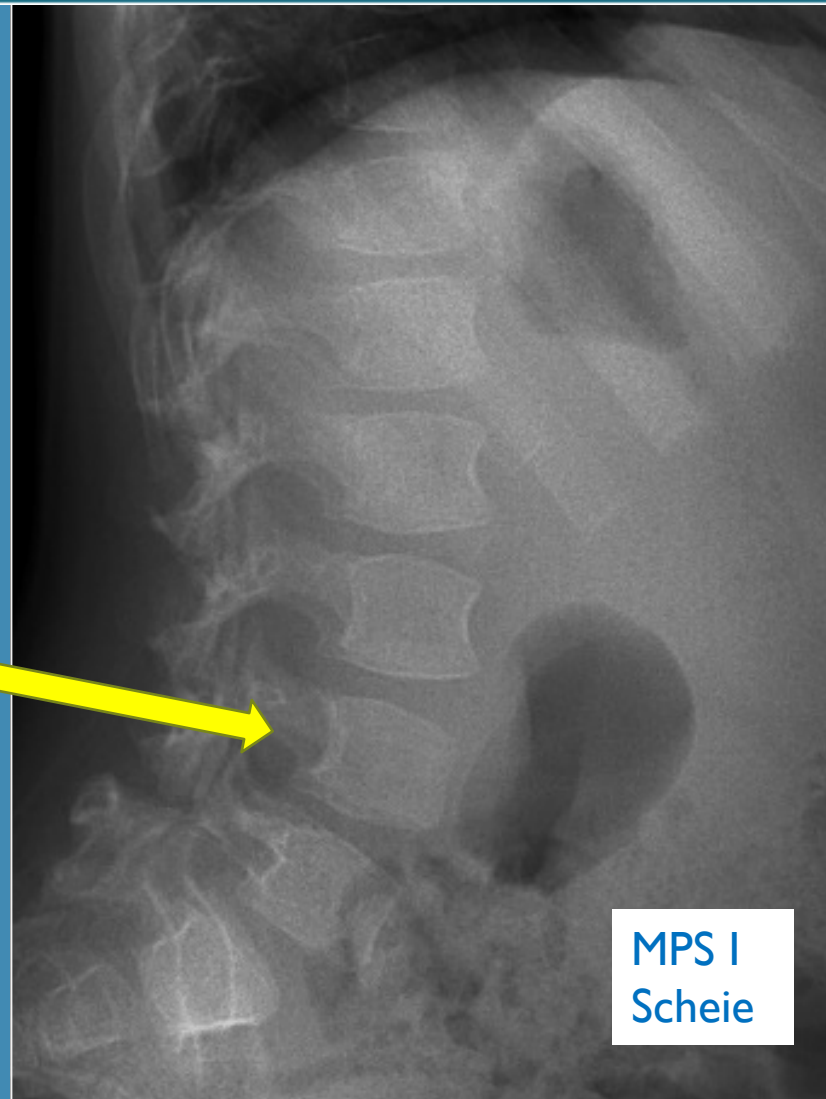
4

couche



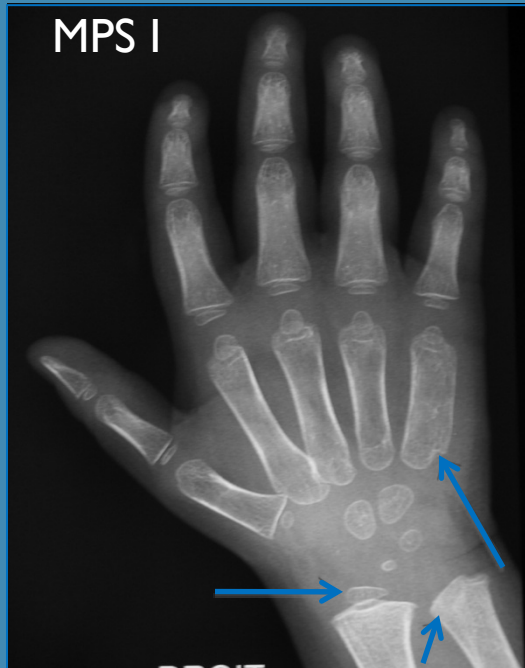


THORACO-LUMBAR SPINE



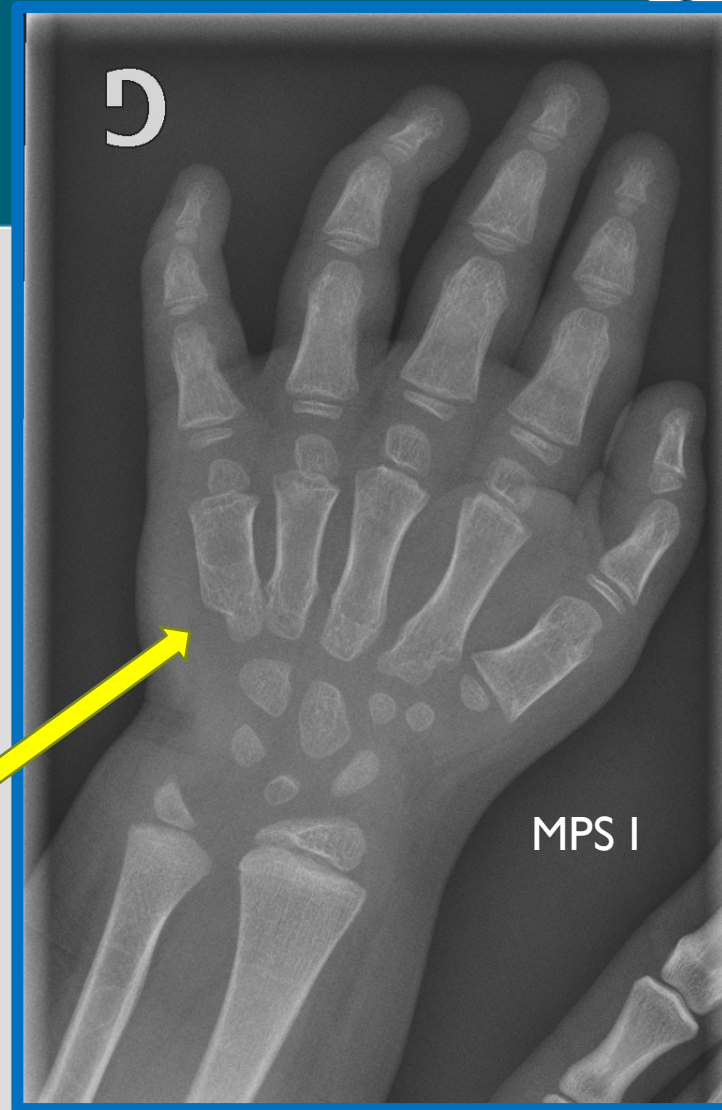


**HAND X-RAY
MULTIPLE BONE LESIONS**



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Notch



Normal

- Small radial epiphysis
- Delayed carpal bone ossification
- Pointed epiphysis, thick and wide metacarpals and phalanges
- First phalanges in cone



Girl, MPS I,
2y 1/2

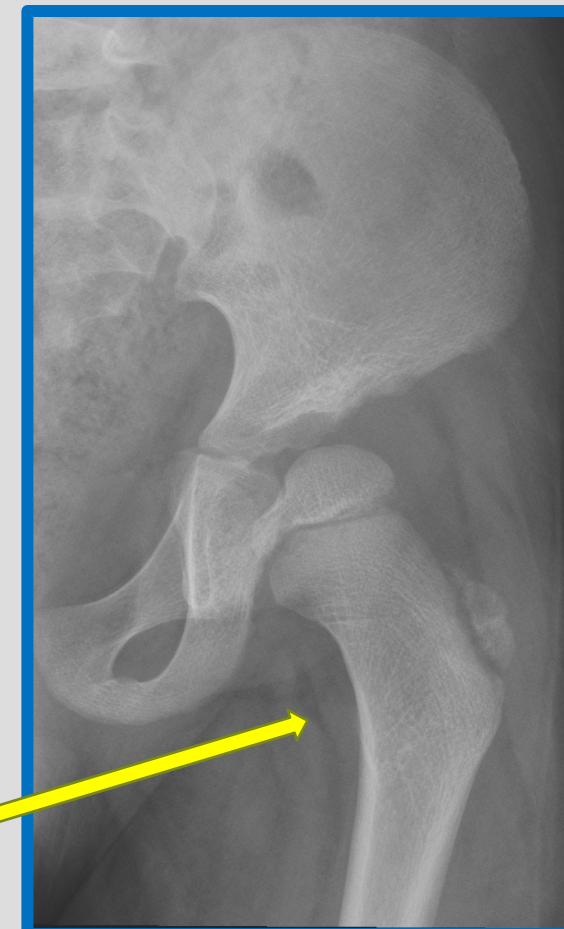
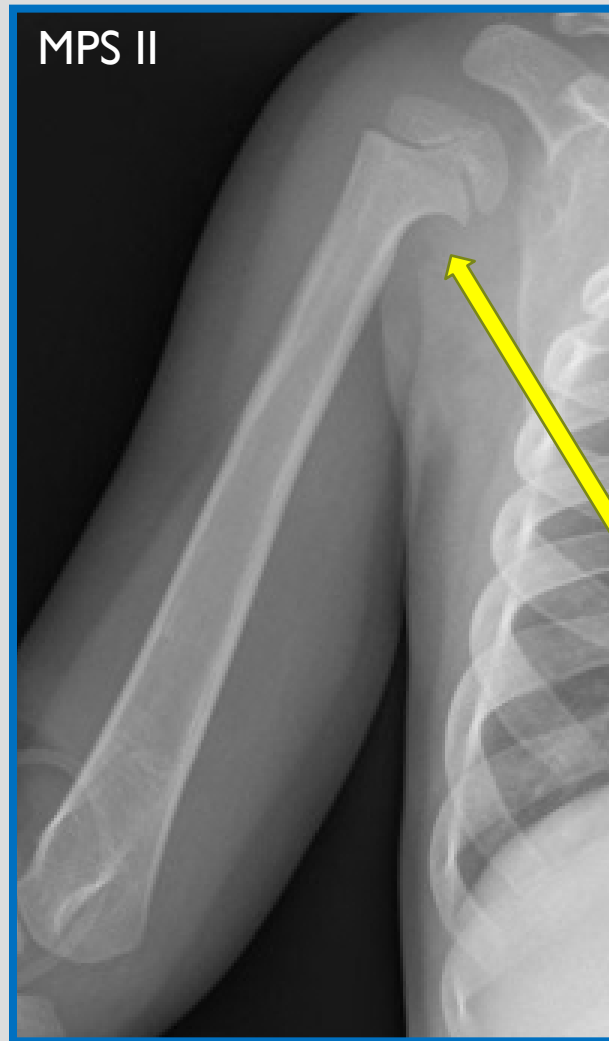




LONG BONES

- Proximal humeral notching
- long and narrow femoral neck

MPS II



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CONCLUSION

« AN INFORMED RADIOLOGIST IS WORTH 2 »

- Skeletal involvement : spotting small signs
- Child diagnosed and managed early = better prognosis



THANK YOU FOR YOUR
ATTENTION

