

How the radiologist can help the physician?

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Disclosures

Consultant for

- ❖ Sanofi
- **❖** Biomarin
- Orthopediatrics
- Orthofix





Enzyme deficiency

Glycosamino-glycanes (GAG) accumulation in lysosomes

Chondroblasts

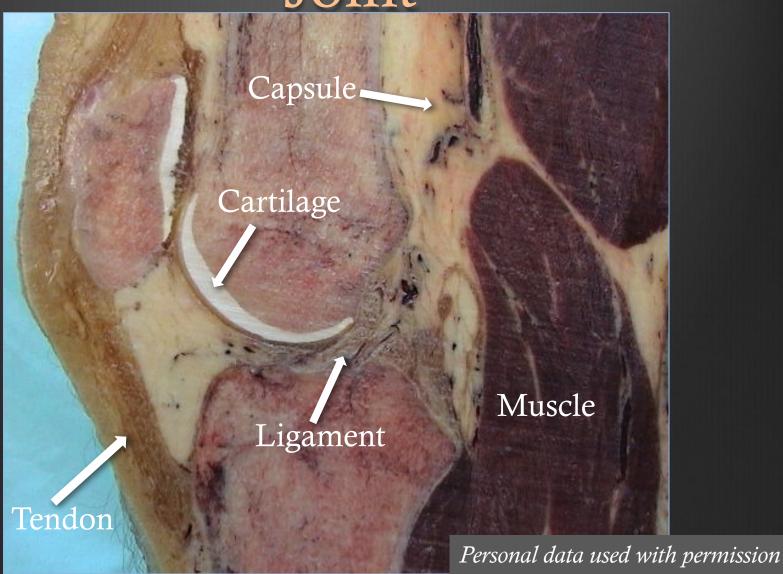
Bone side effects

Fibroblasts

connective soft tissues



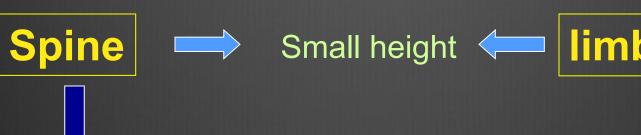
Joint







Bone side effects



T12, L1 deforrmity

Kyphosis

Odontoideum hypoplasia

Spinal canal narrowing

Cervical spine instability

Myelopathy



Hips dysplasia

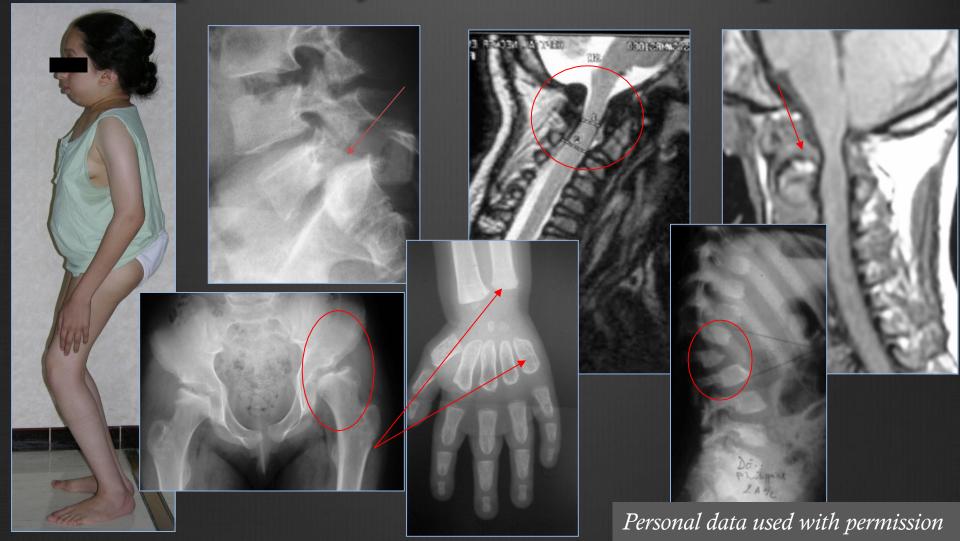
Genu valgum

Hand deformities

Joints stiffness



Typical dysostosis multiplex





But in some clinical cases...





















What is their common point?...



Absence of face dysmophy....

However....











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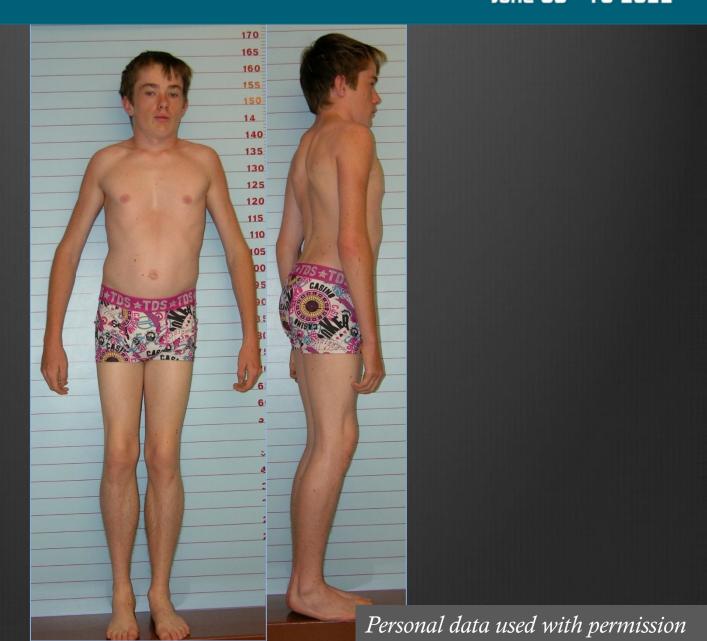
Physical examination...





















What is their common point?...



Limited joint mobility and short stature...

However...



... There is sometimes « hyperlaxity»...





And don't forget the others physical signs...



AP deviation of lower limbs

- ⊕Genu valgum
 - & Proximal part of tibia
 - Femur less often
- MPS I, IV and VI







Carpal tunnel syndrome



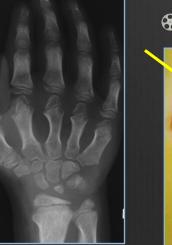
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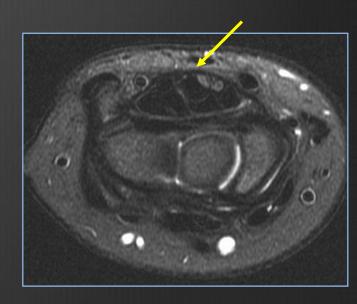
Carpal tunnel syndrome

Mechanisms

- Height decrease of the wrist
- Carpal ligament thickeness
- Synovitis hypertrophy



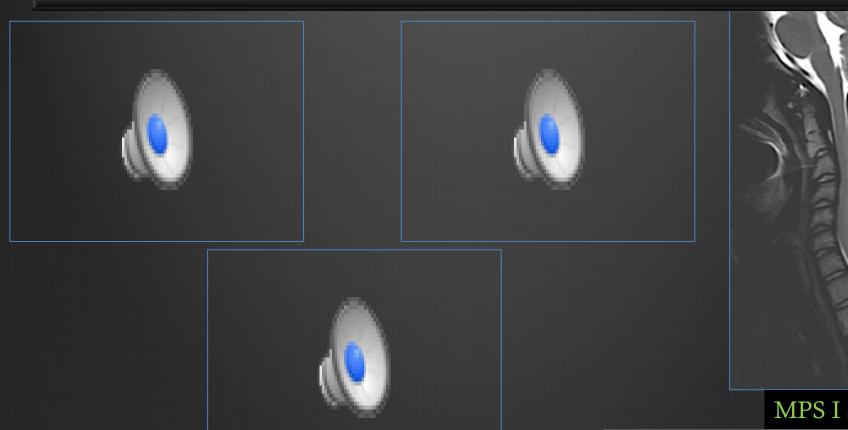




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Full neurological examination





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Why the X-rays are crucial?

What are the essential X-rays?





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