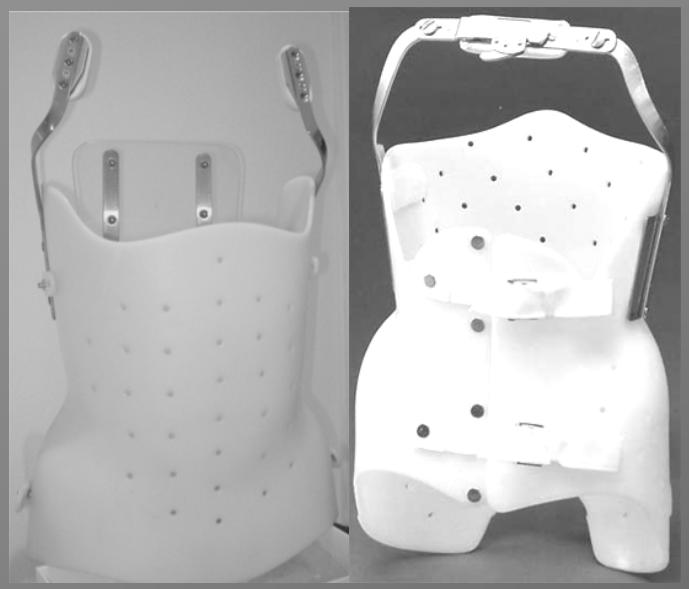
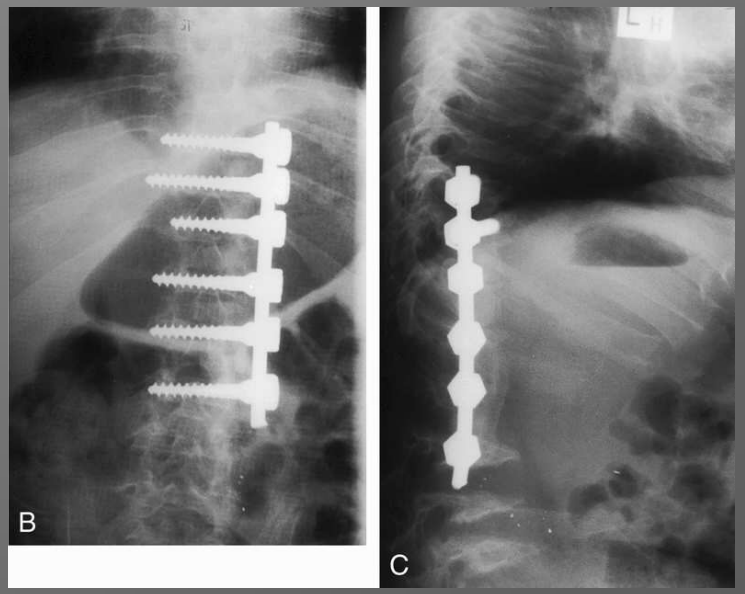
MPS: Spine involvement focus on hyphosis of thoracolumbar spine





- Anti-gravity bracing
- Surgical strumental stabilization

Form: Tandon et al. Spinal Problems in MPS I (Hurler Syndrome) JBJS Br,78-B(6),nov 1996.938-944

MPS: Spine involvement focus on cervical upper spine

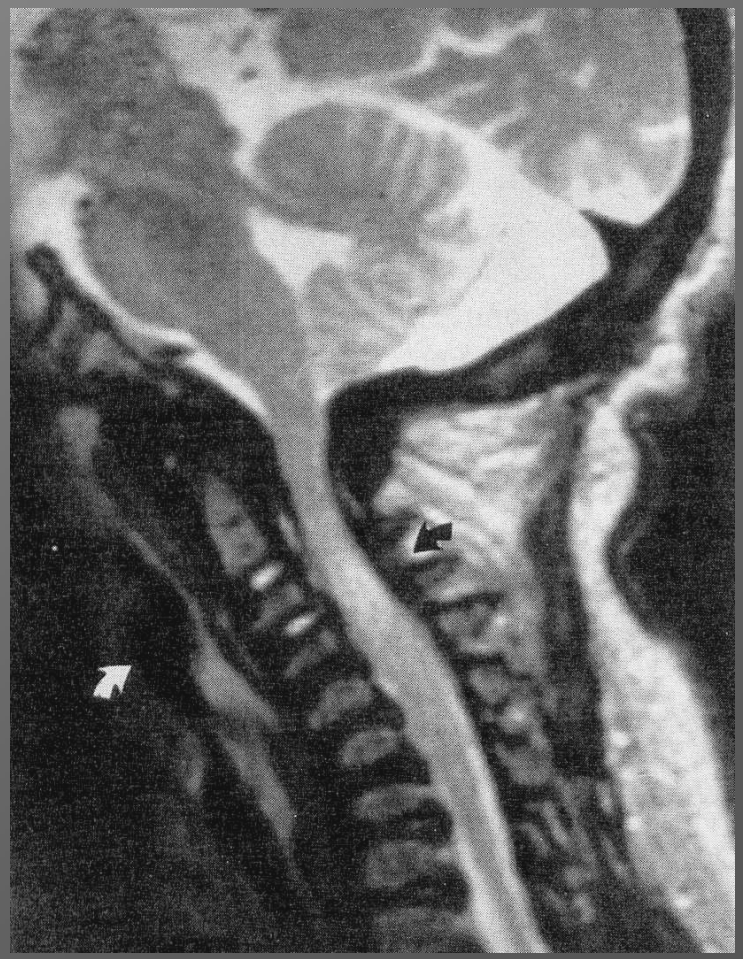
- Joint instability
  - of the upper cervical spine
  - of the lover cervical spine
- GAGs periarticular storage
- Combination of both

MPS: Spine involvement focus on cervical upper spine

Glycosaminoglycan periarticular storage

Sagittal image of the cervical spine:

- retroodontoid soft tissue storage leading to cord compression
- High signal changes in the cord (suggestive for local oedema)



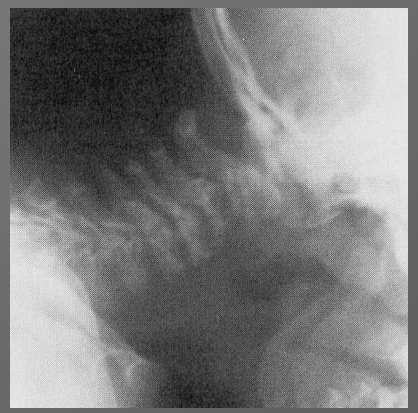
Form: SL Wienstein.The Pediatric Spine. LWW. 2001.

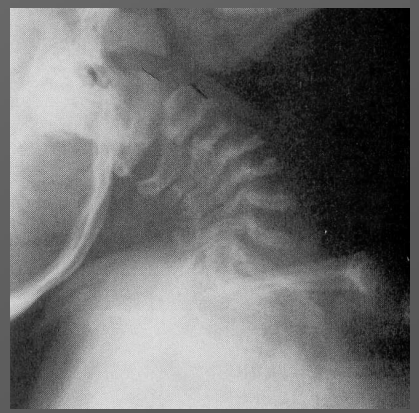
MPS: Spine involvement focus on cervical upper spine

Instability of the upper cervical spine

Dynamic image of the cervical spine in a 3-year-old with Morquio's syndrome:

- Flexion-extension radiographs demonstrate the C1-C2 instability secondary to hypoplasia of the odontoid



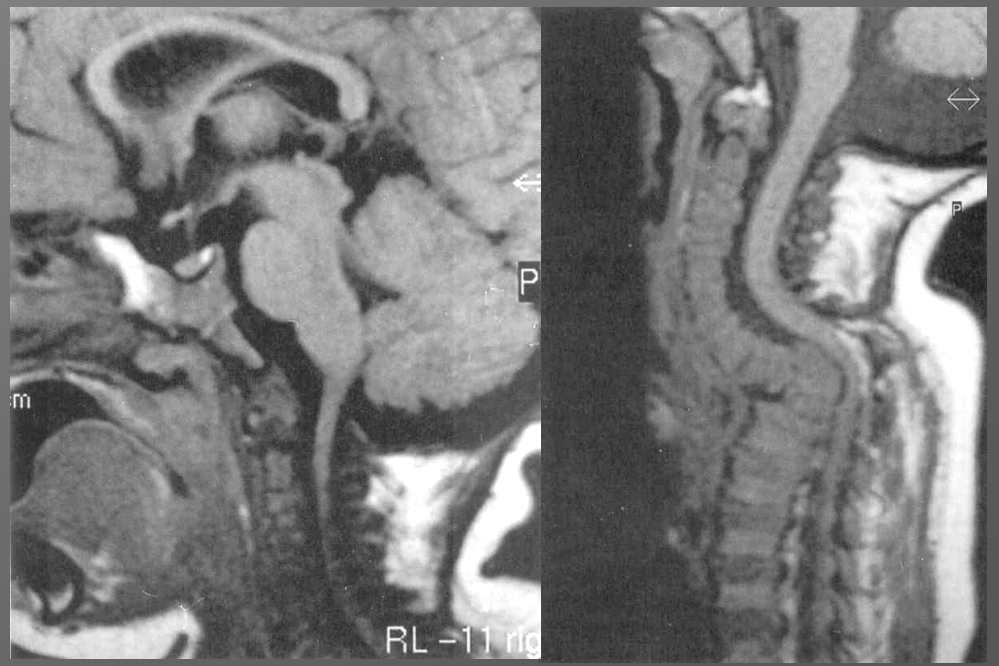


Form: SL Wienstein.The Pediatric Spine. LWW. 2001.

MPS: Spine involvement focus on cervical upper spine

Instability of the lower cervical spine

From: Khan at al. Spine 2003

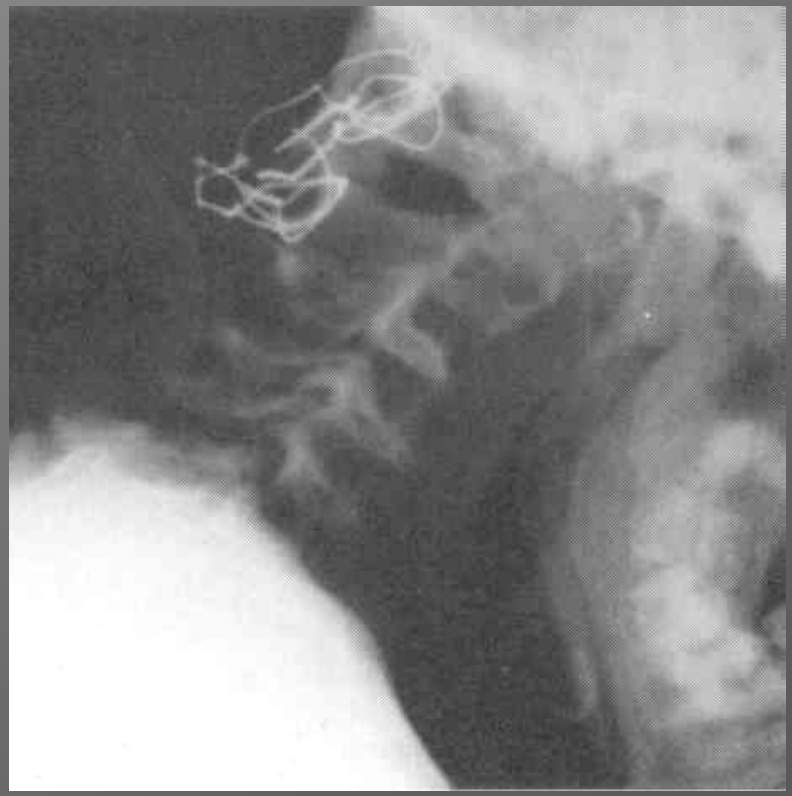


From: Tandon et al. Spinal Problems in MPS I (Hurler Syndrome) JBJS Br,78-B(6),nov 1996.938-944

MPS: Spine involvement focus on cervical upper spine

C1-C2 Cervical subluxation:

Postoperative radiograph showing posterior occipitocervical fusion and an ossified odontoid peg.



From: Ransford: J Bone Joint Surg Br, Volume 78-B(2).March 1996.307-313

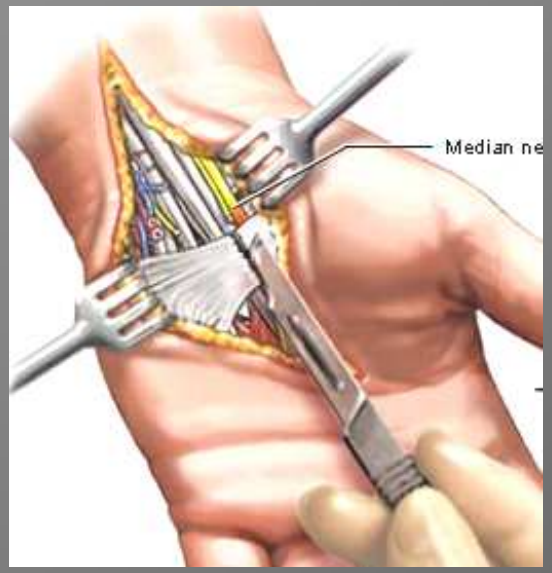
MPS: Upper Limbs involvement

- Stiffness of shoulder, elbow and wrist

GG,f,3 ½ years. MPS VI

MPS: Upper Limbs involvement

- Carpal tunnel syndrome



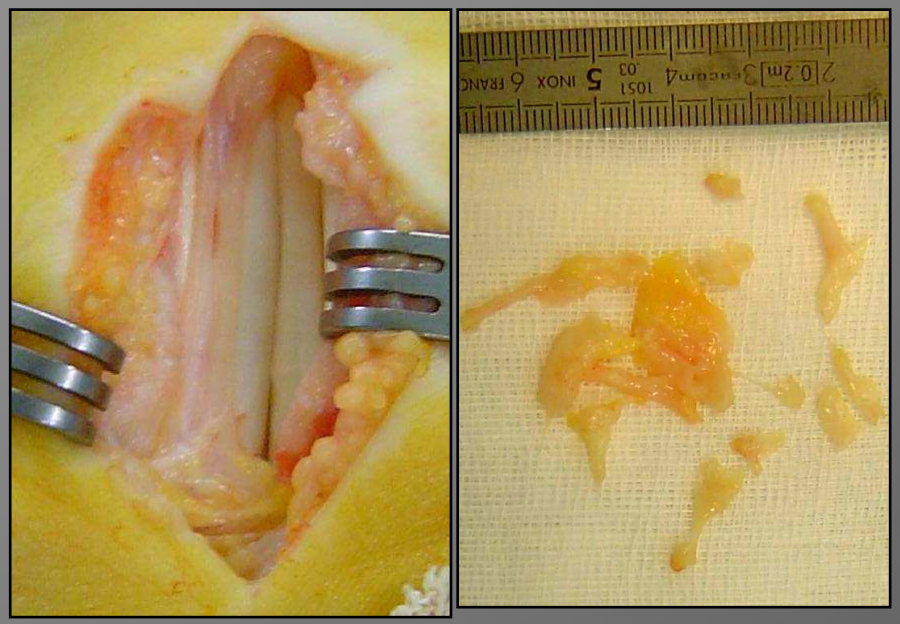
Cause	Number of cases	Studies
Mucopolysaccharidosis	95	[4–13]
Mucolipidosis	22	[6,12,13,14–19]
Familial CTS	11	[3,5,15,20–24]
HNPP	1	[5]
Schwartz–Jampel syndrome	1	[5]
Melorheostosis and Leri's pleonosteosis	3	[2,14,25]
Dejerine–Sottas syndrome	1	[14]
Weil–Marchesani syndrome	1	[14]
Idiopathic	4	[15,26–28]
Sports-related	6	[5,15,23]
Fibrolipomatous hamartoma/NTOM	4	[14,29,30]
Intraneural perineuroma	1	[31]
Haemangioma of median nerve	1	[32]
Musculotendinous malformation	2	[14,33]
Trigger finger	1	[5]
Klippel–Trénaunay syndrome	1	[34]
Poland syndrome	2	[33,35]
Scleroderma	1	[15]
Trauma	10	[15,33,36–38]
Total	163	

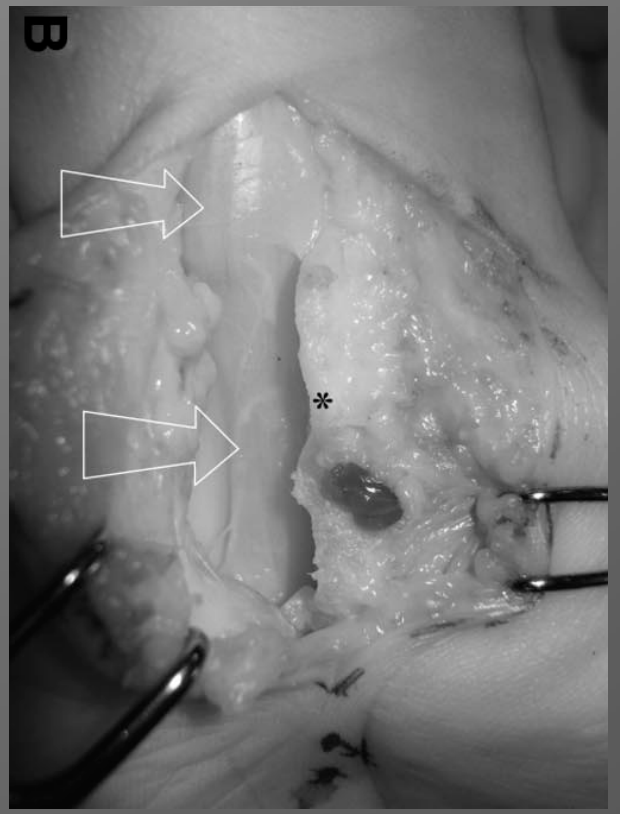
CTS, carpal tunnel syndrome; HNPP, hereditary neuropathy with liability to pressure palsies; NTOM, nerve territory-oriented macrodactyly.

Form: Van Meir. Carpal Tunnel Syndrome in Children JPO Jan 2005

MPS: Upper Limbs involvement

- Carpal tunnel syndrome
  - Glycosaminoglycans storage





5 y, MPS III

From: Haddad: J Bone Joint Surg Br, Vol. 79-B(4). 1997

MPS: Upper Limbs involvement

- Trigger finger
  - Glycosaminoglycans storage

AY, 3 yy. MPS I

Hip &MPS: general view




- loss of sphericity of the femoral head
- Flat acetabula
- Increased neck-diaphyseal angle
- Migration of the hip

Hip &MPS: general view

- MPS I, M,
- Annual X-ray of the same patient
  - Migration
  - Loss of sphericity
  - Flat acetabula
  - Neck-diaphyseal angle

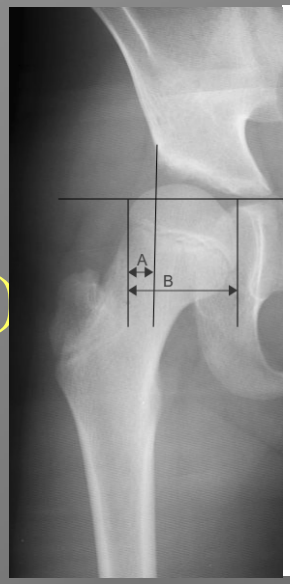
Ashby E, Baker M, Eastwood D. J Pediatr Orthop 2015

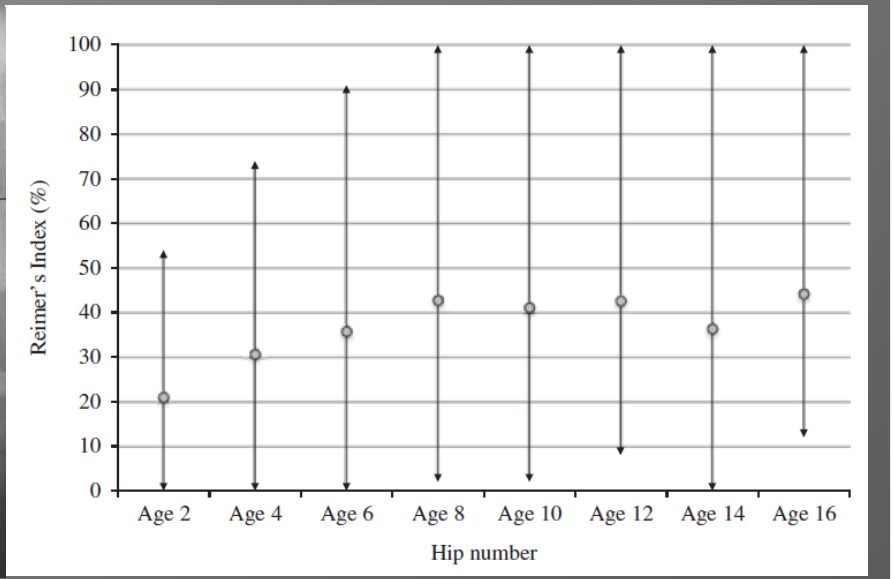
Hip & MPS: natural history

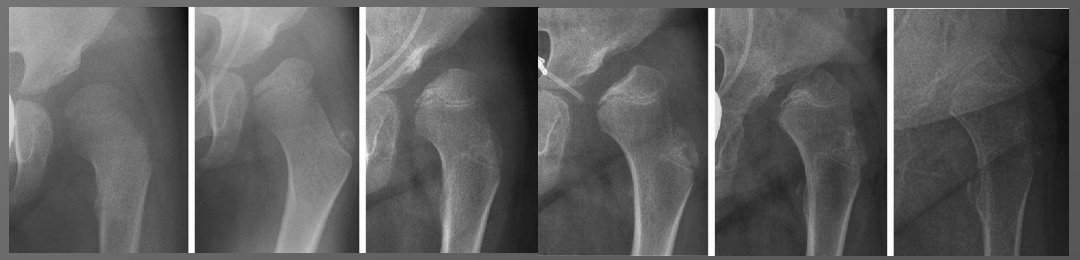
- Miration
- 64 hips (MPS I & II)

Reimar's Index
(= A / B * 100)

RI increases between 2 and 8 yy







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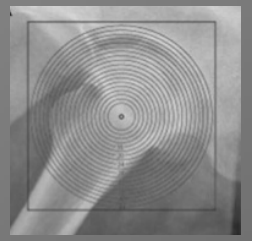
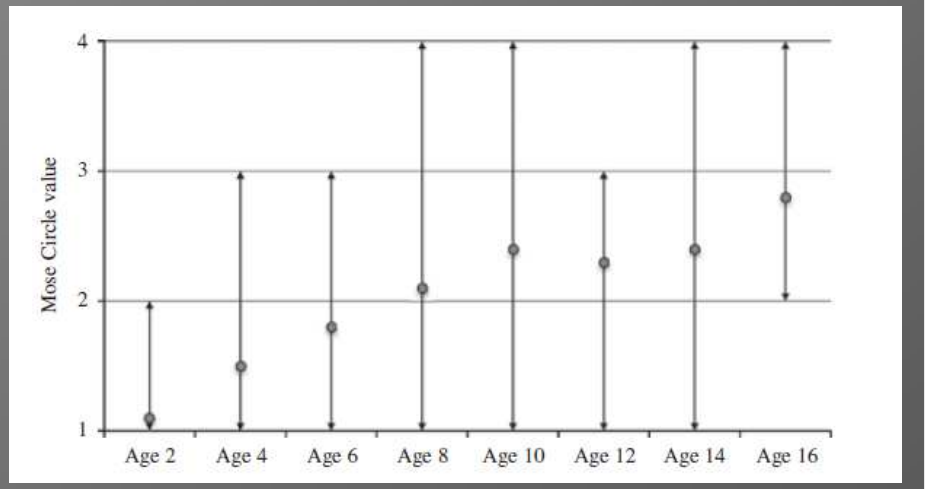
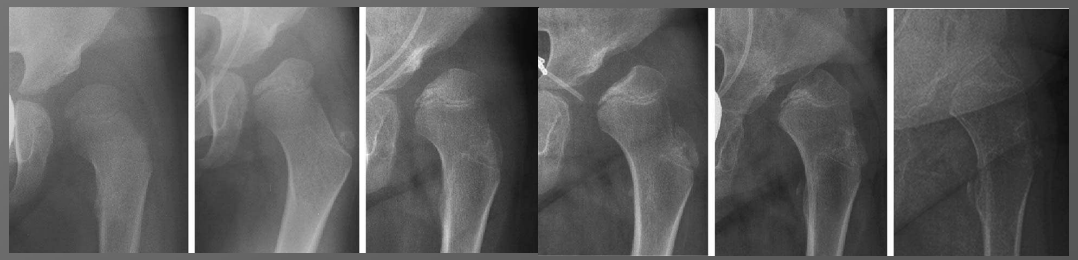
Hip & MPS: natural history

- Loss of sphericity
- Hip & MPS: natural history





Mose technique



Loss of spericity betwee 3 and 8 yy

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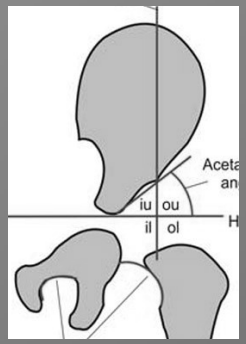
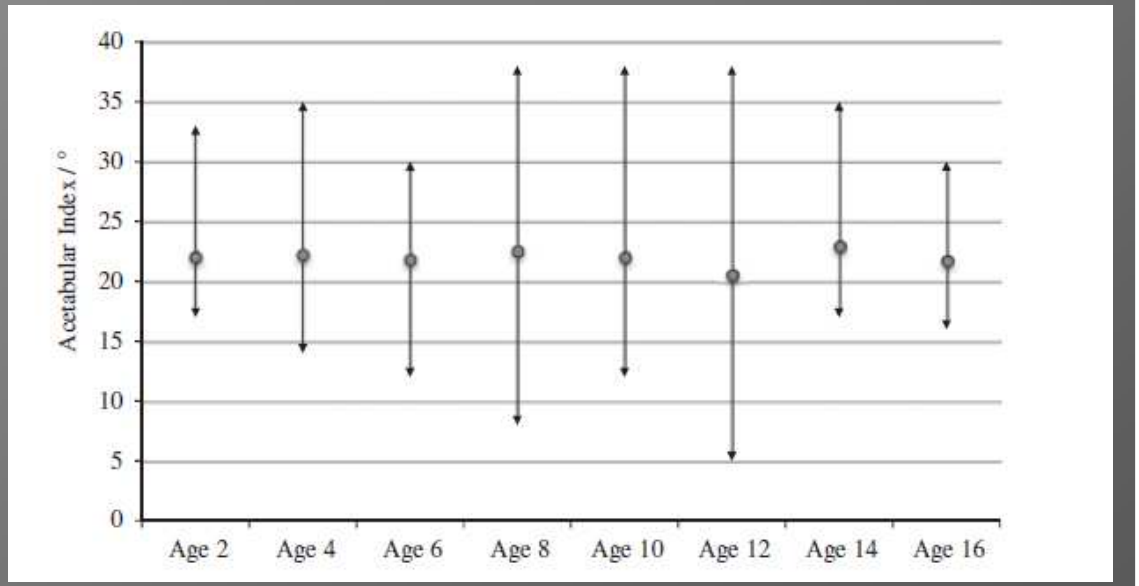
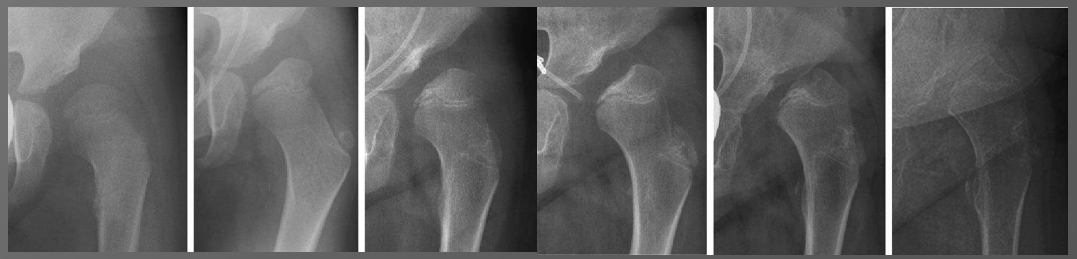
Hip & MPS: natural history

- Acetabular Index
- Hip & MPS: natural history



- We do not have the physiological decrease between the 2 and the 6 yy





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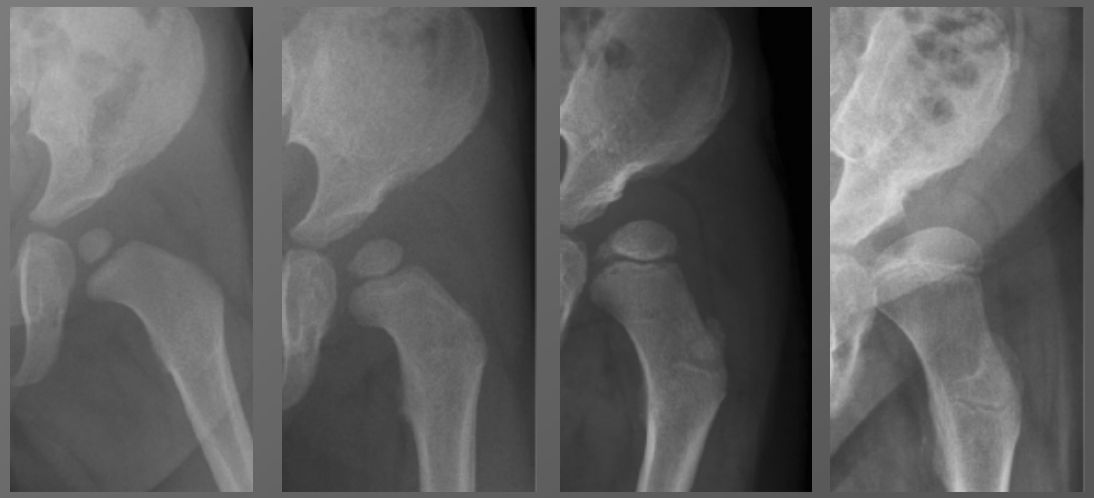
Hip & MPS: natural history

- Neck-diaphyseal angle
  - Augmented (Described by several Authors but not exactly quantified)



Langereis E, Borgo A et al. Orphanet Jour, 2013

Hip & MPS: natural history

- MPS III
  - 50 % of patients have a involvement of the femoral head
  - Generally the acetabula is less flat
  - High prevalence of necrosis of the femoral head (after age 10)
  - (generally very poor general conditions)

13 yy

17 yy

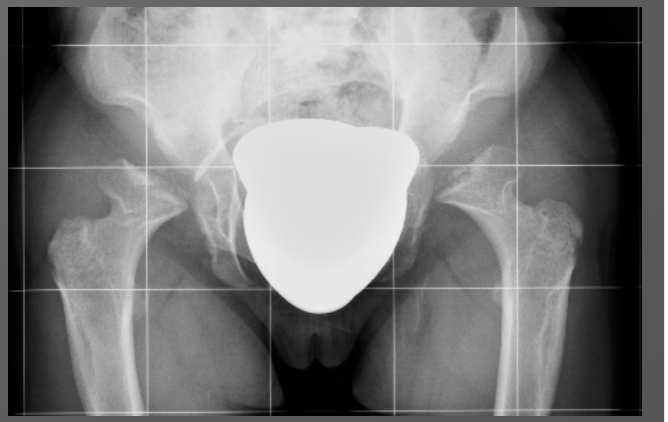
18 yy

White K, Karol LA et al. J Pediatr Orthop, 2011
De Ruijter J, Maas M et al. Mol Gen and Metab, 2013

Hip & MPS: natural history

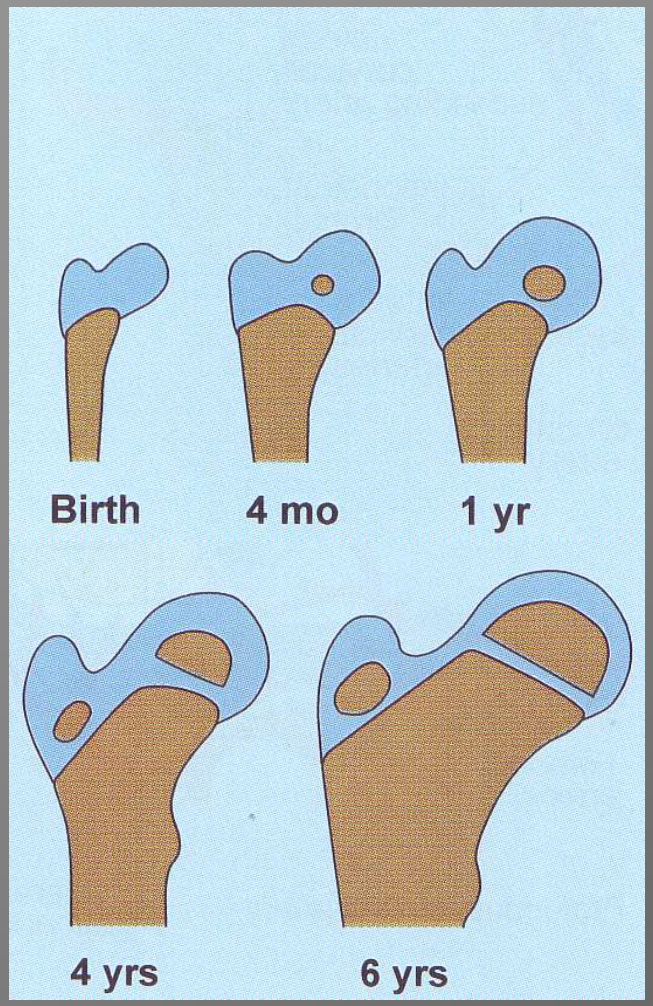
- MPS VI
  - Hip always involved
  - Generally symptoms are less severe
  - Generally low tendency to migration
  - (lack of literature)
  - (role of surgery not clear)

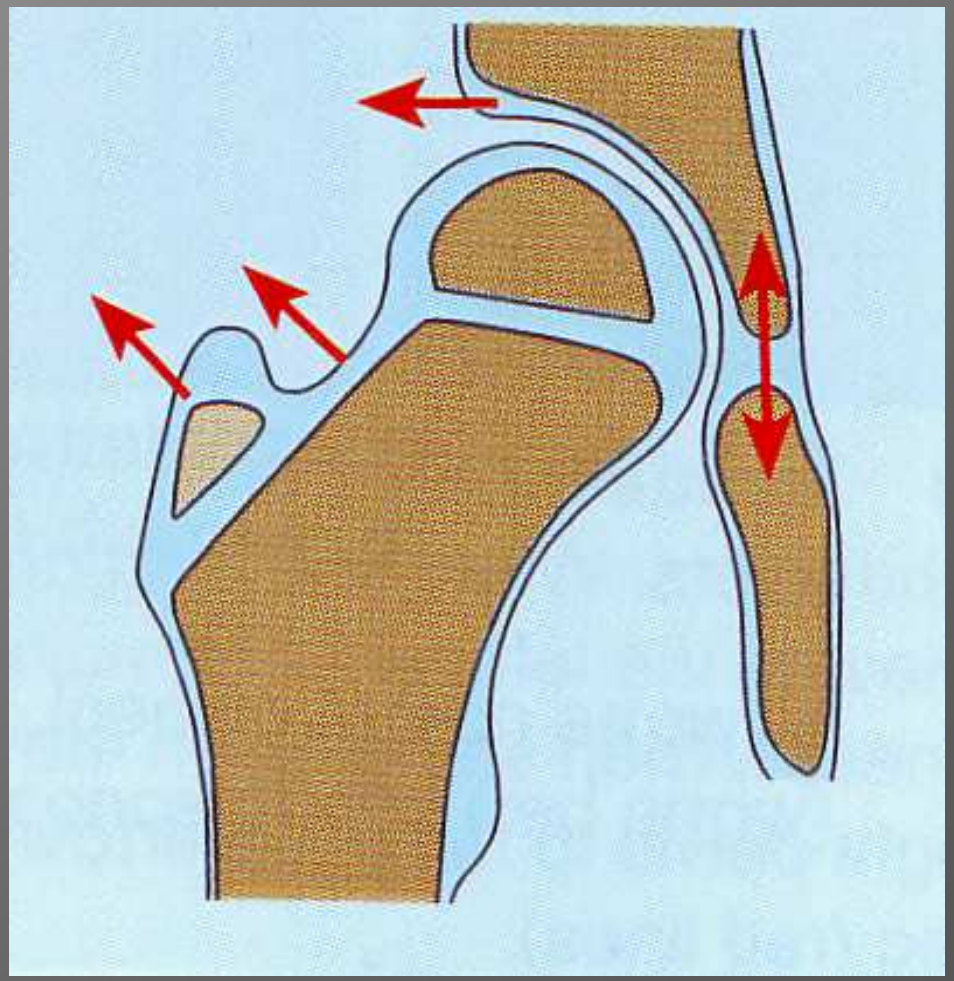
6 anni



15 anni

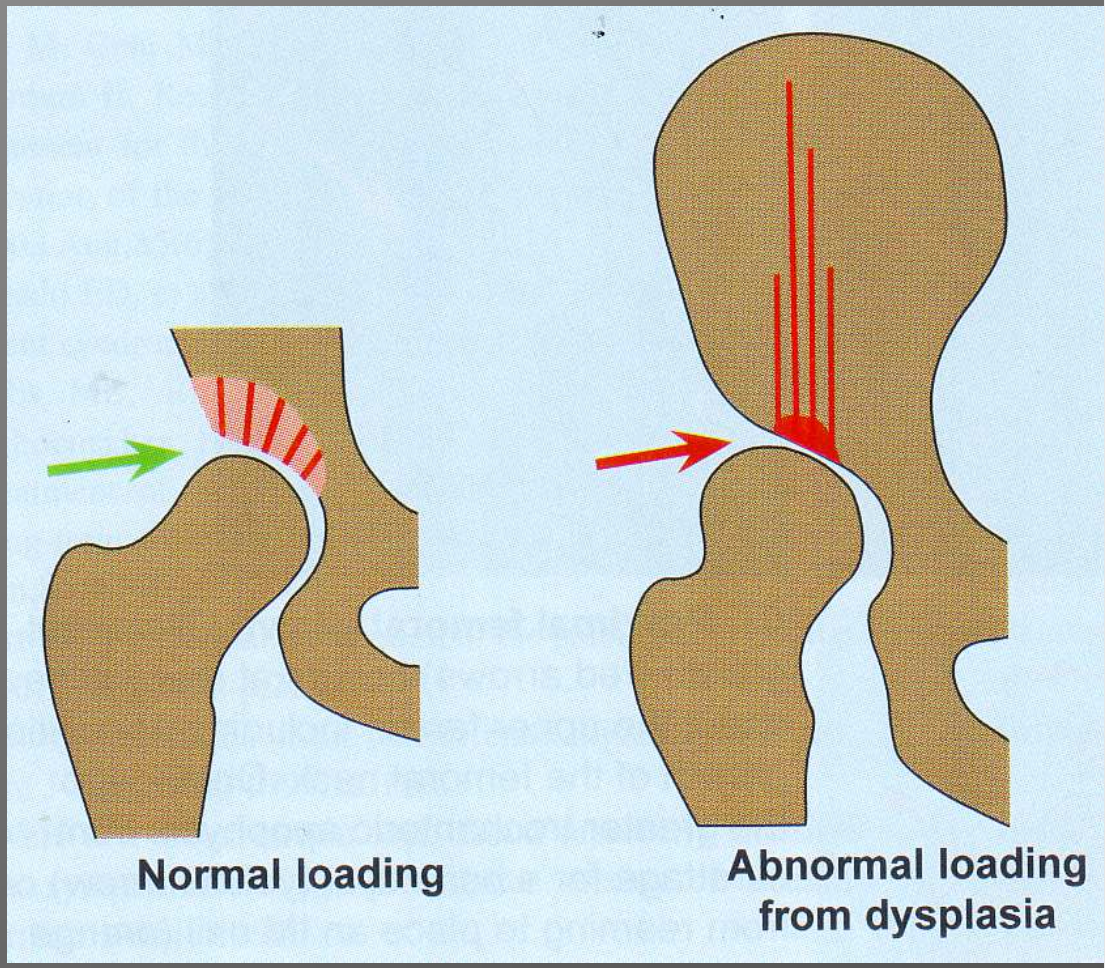
Physiological hip development





Tonnis 1984 and Staheli 2008

Physiological hip development



Tonnis 1984 and Staheli 2008