Advances in Achondroplasia 2023

21–22 April 2023, Frankfurt

Medical Faculty Mannheim
Heidelberg University

University Medical Centre Mannheim

Developments in Achondroplasia – Spring 2023
Following up Adolescents and Adults with Achondroplasia: Is it Necessary?

Spinal Issues in Adolescents and Adults with Achondroplasia

Philip Kunkel

Pediatric Neurosurgery
Department of Neurosurgery, University Hospital Mannheim, Germany

EAF
European Achondroplasia Forum

UMM
UNIVERSITÄTSMEDIZIN MANNHEIM

Medical Faculty Mannheim
Heidelberg University

University Medical Centre Mannheim

Disclosure

Conflicts of interest:

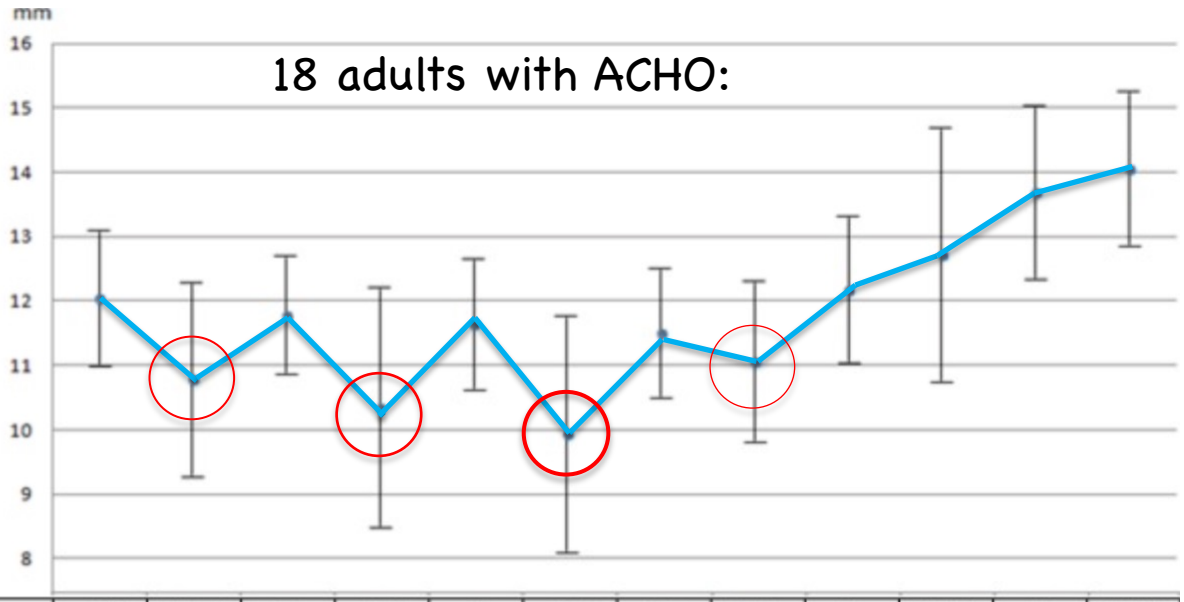
BIOMARIN® - Travel expenses

Medical Faculty Mannheim
Heidelberg University
University Medical Centre Mannheim

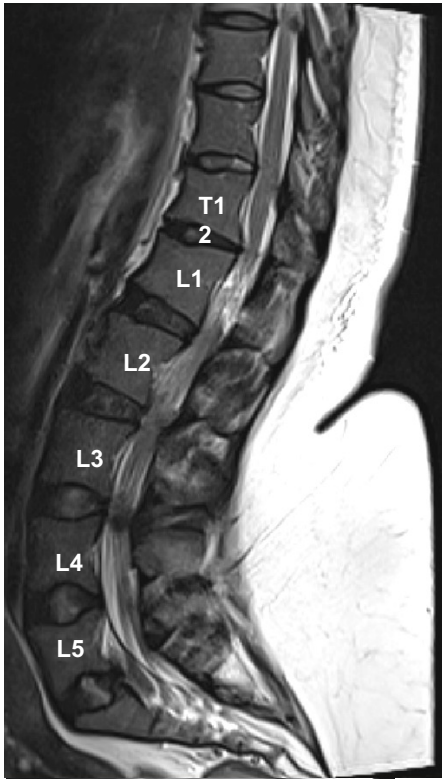
1. Thoraco-lumbar and lumbar spinal stenosis
2. Thoraco-lumbar kyphosis
3. Spinal stenosis AND thoraco-lumbar kyphosis
4. Spinal stenosis cervical / not CCJ

Lumbar spinal stenosis

Medical Faculty Mannheim
Heidelberg University
University Medical Centre Mannheim

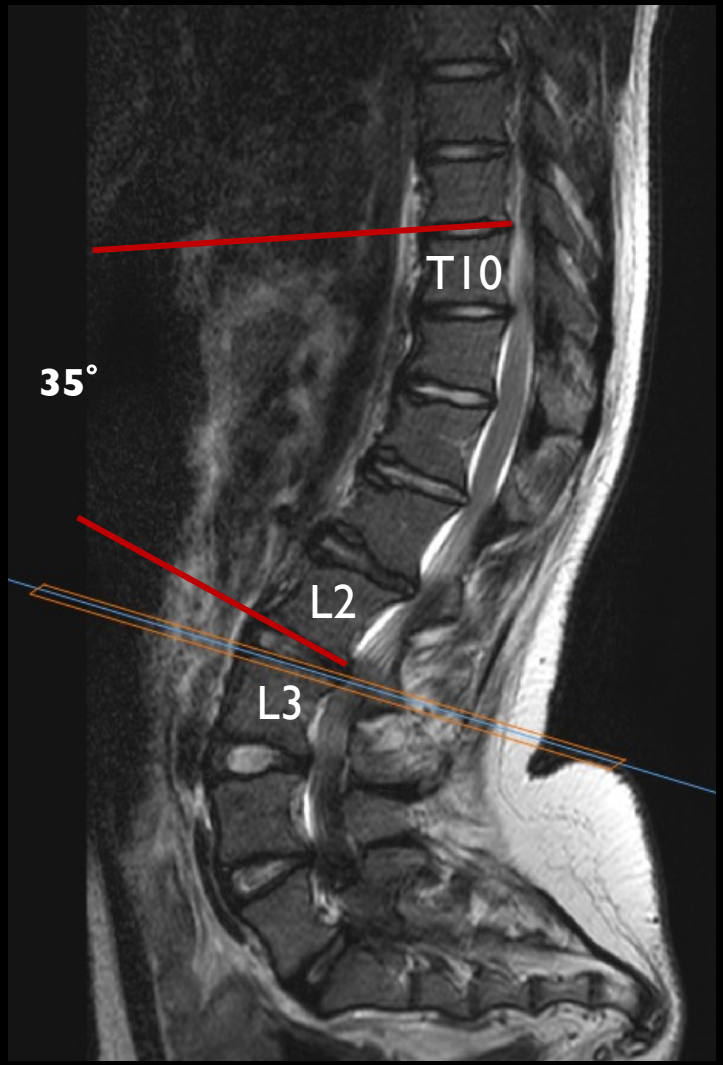


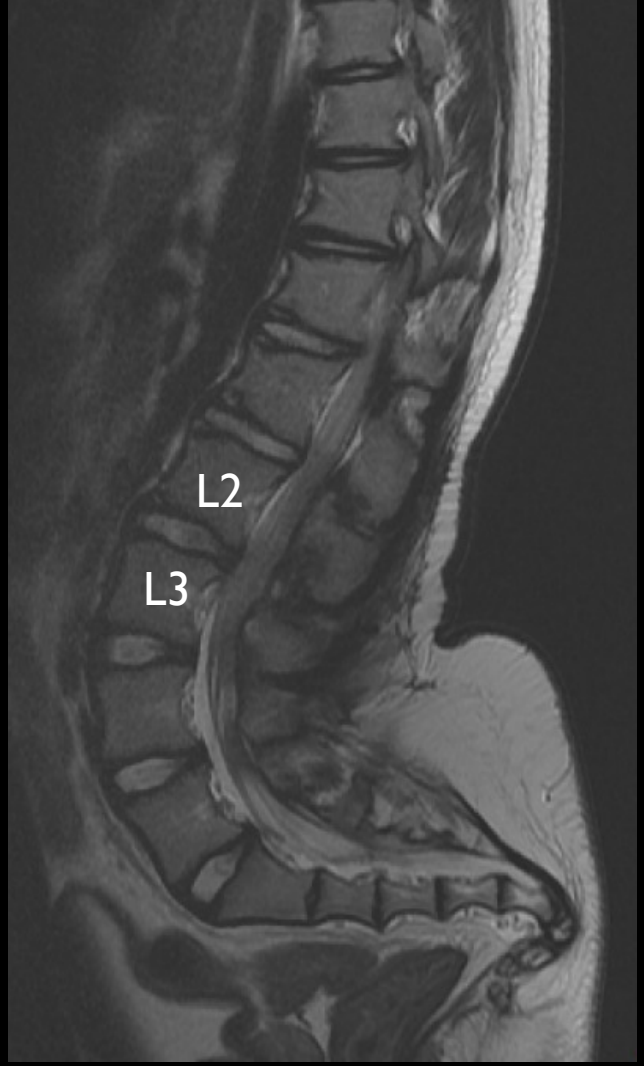
Spinal levels (n)	T12 (18)	T12-L1 (17)	L1 (17)	L1-L2 (17)	L2 (17)	L2-L3 (16)	L3 (16)	L3-L4 (16)	L4 (16)	L4-L5 (16)	L5 (16)	L5-S1 (18)
Antero-posterior diameter, mm (SD)	12 (1)	10.8 (1.5)	11.8 (0.9)	10.3 (1.8)	11.6 (1)	9.9 (1.8)	11.5 (1)	11 (1.2)	12.1 (1.1)	12.7 (1.9	13.7 (1.3)	14 (1.2)



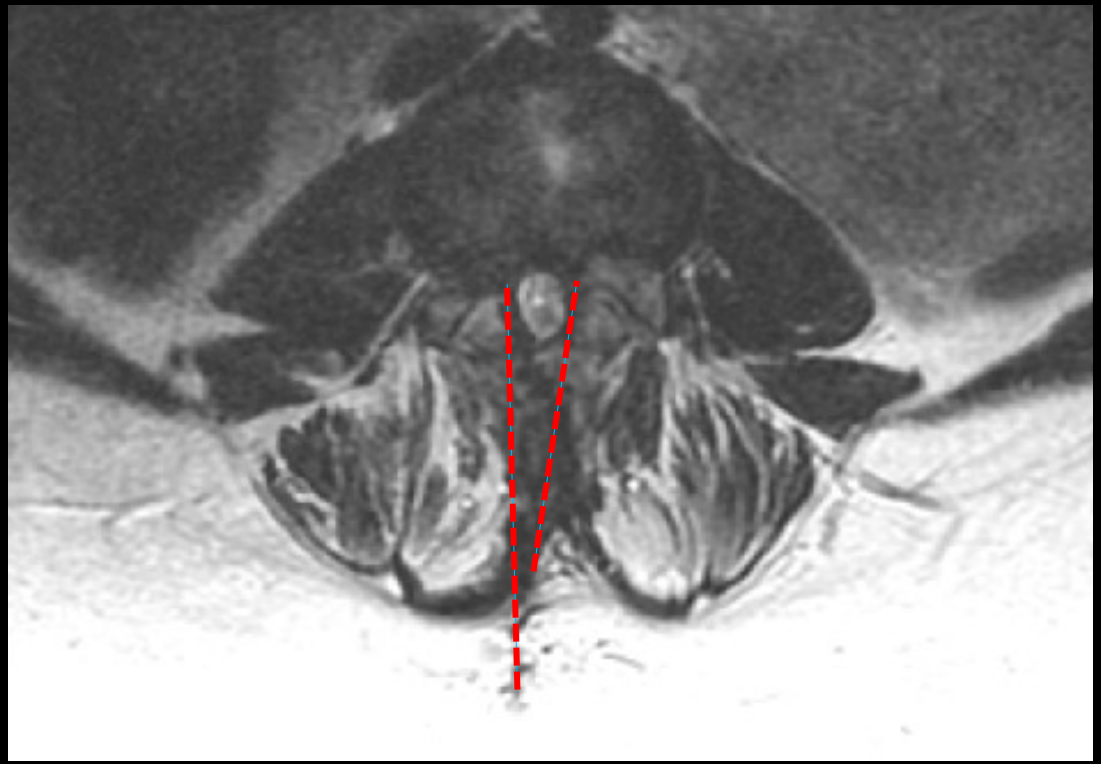
Huet T. et al.: Lumbar spinal stenosis and disc alterations affect the upper lumbar spine in adults with achondroplasia. Sci Rep. 2020

14y/o boy, walk distance 200m

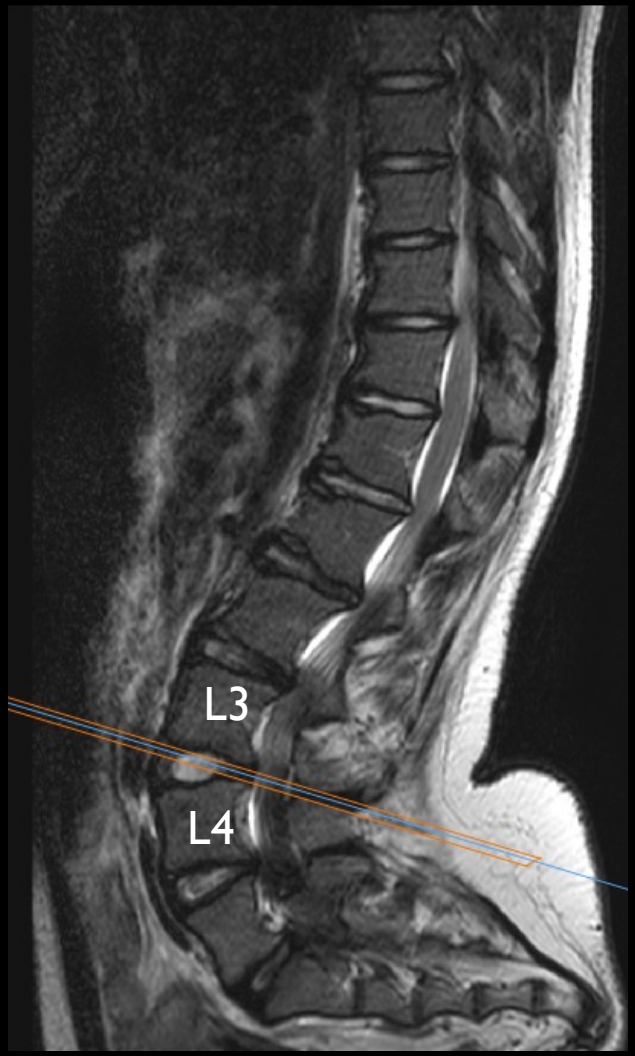


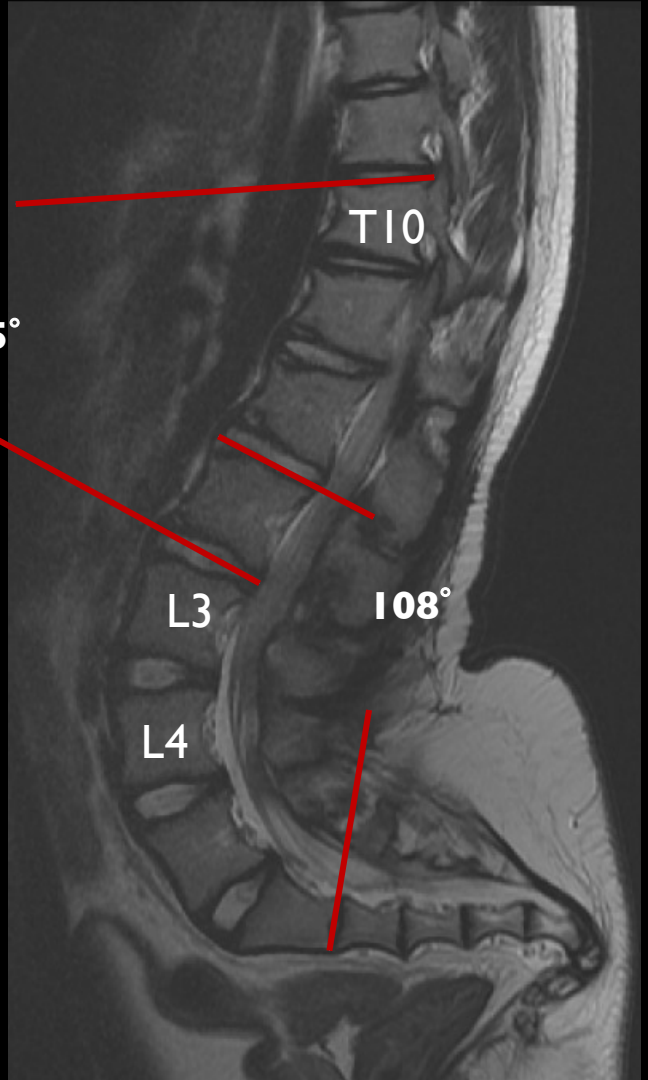


post decompression

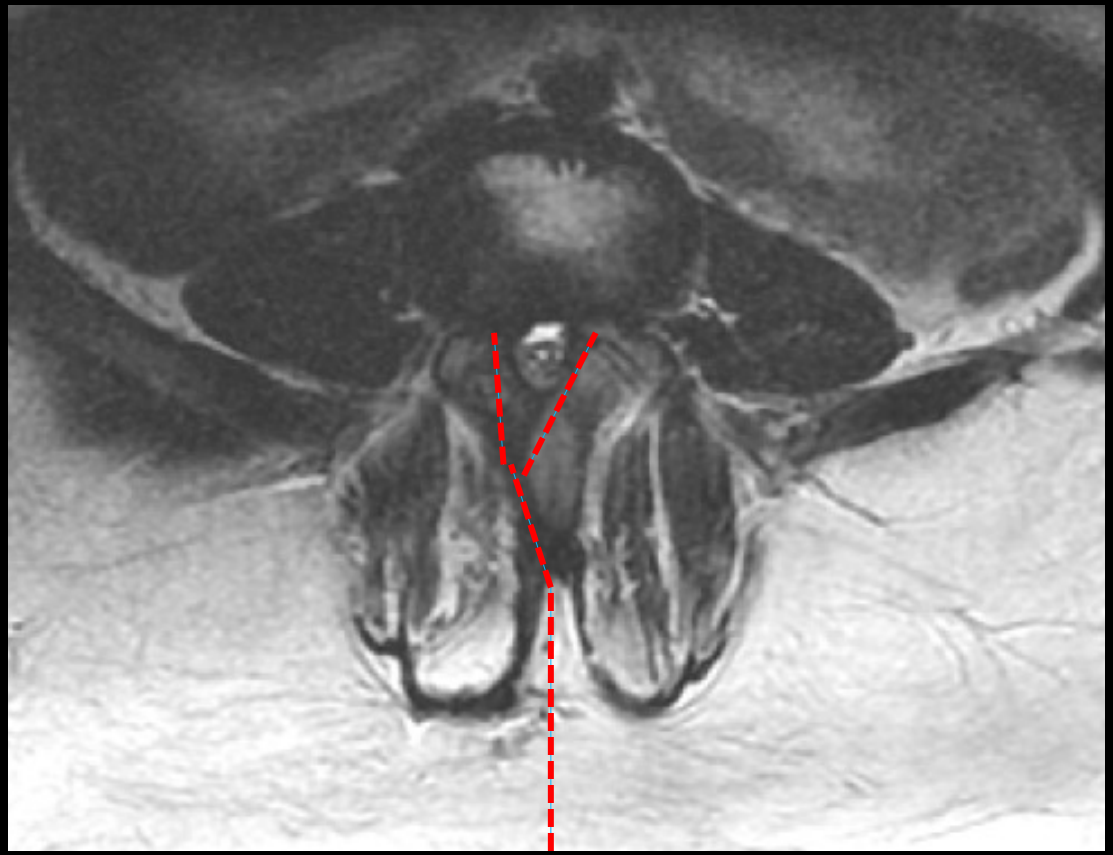


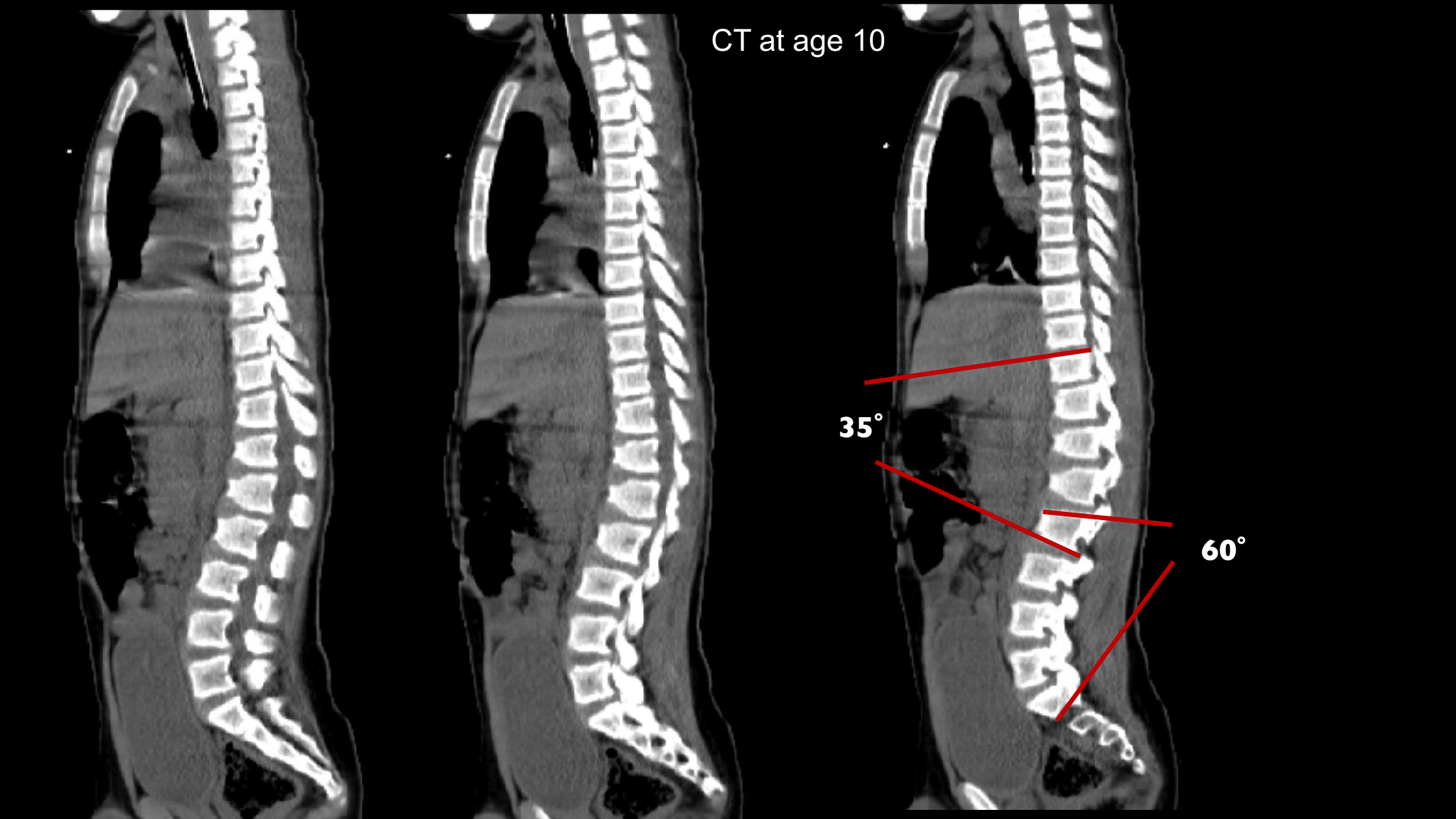
14y/o boy, walk distance 200m





post decompression

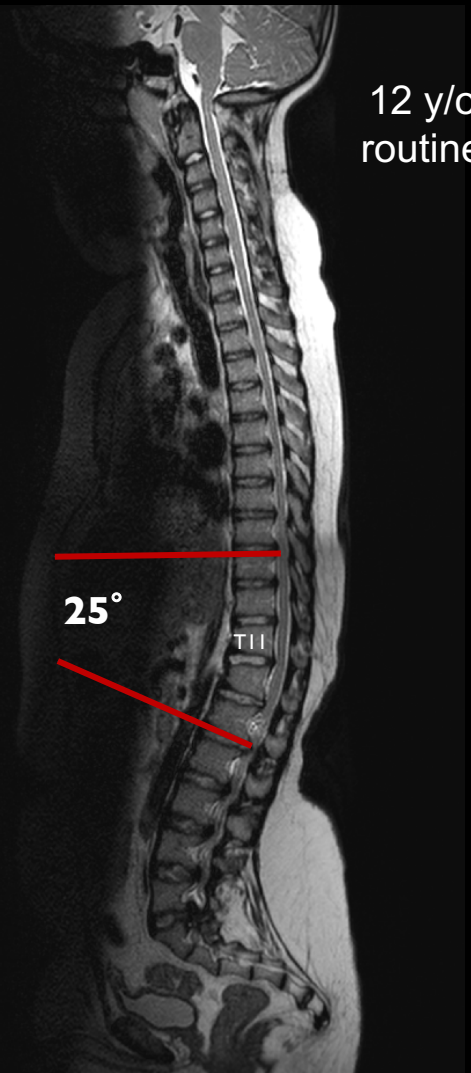



CT at age 10
35°
60°

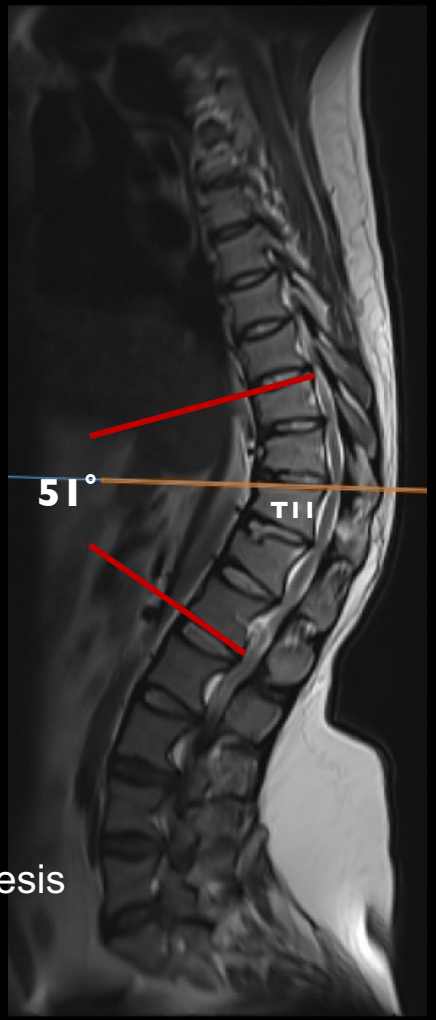
Results of surgery

- 20 adult ACHO patients (age 53 +/- 15 y.), 2007-17, HSS New York
- Most frequent surgery: Thoraco-lumbar multilevel decompression w/o fusion in 57%
- Complications:
  - 36% dural tear
  - 12% neurological complication
  - 8% infection
  - 36% second surgery: 12% additional level decompression, 12% revision, 12% fusion

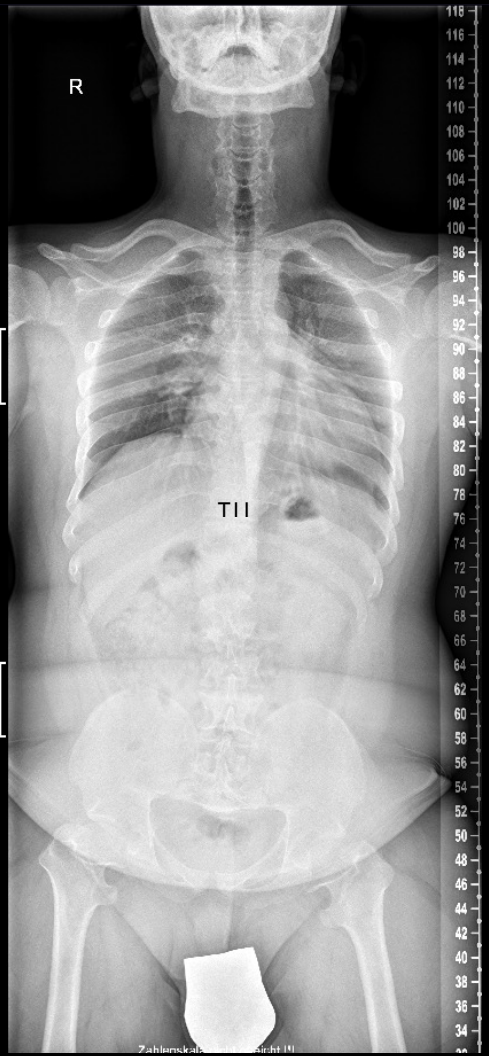
Shafi K. et al.: Complications and revisions after spine surgery in patients with skeletal dysplasia: Have we improved? Global Spine J. 2021

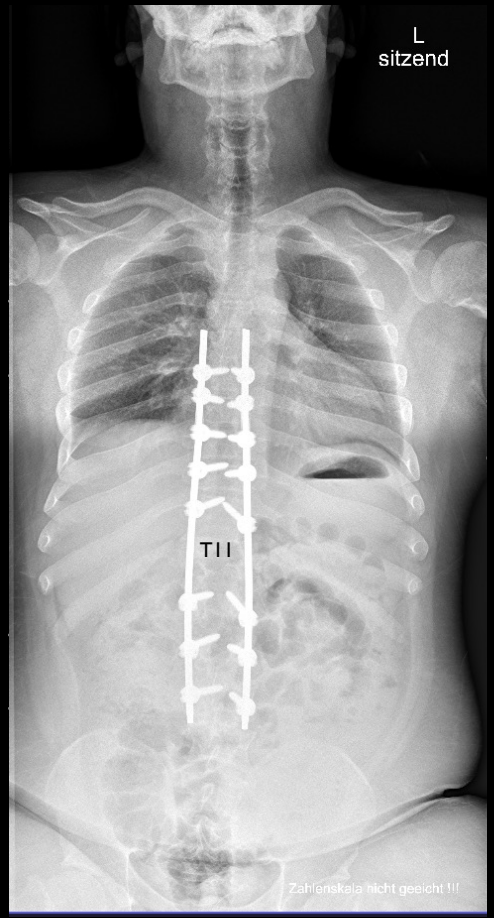


12 y/o boy,
routine MRI

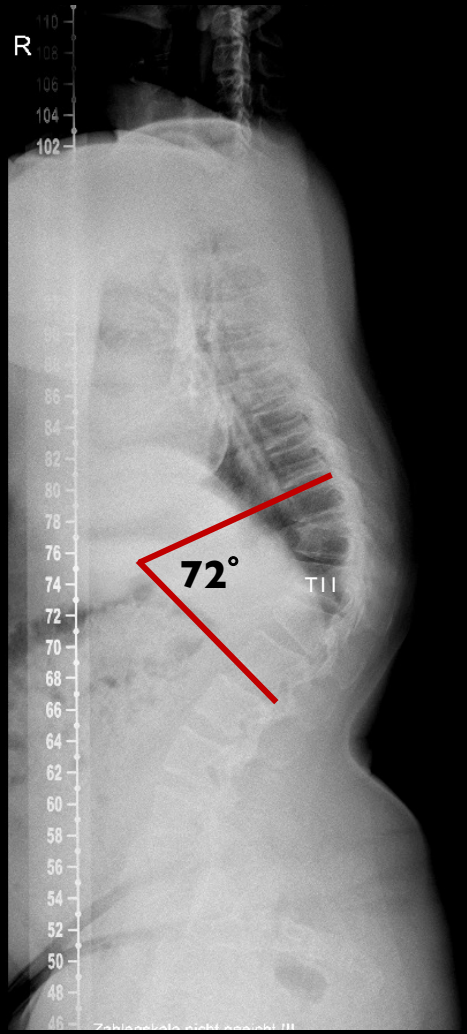


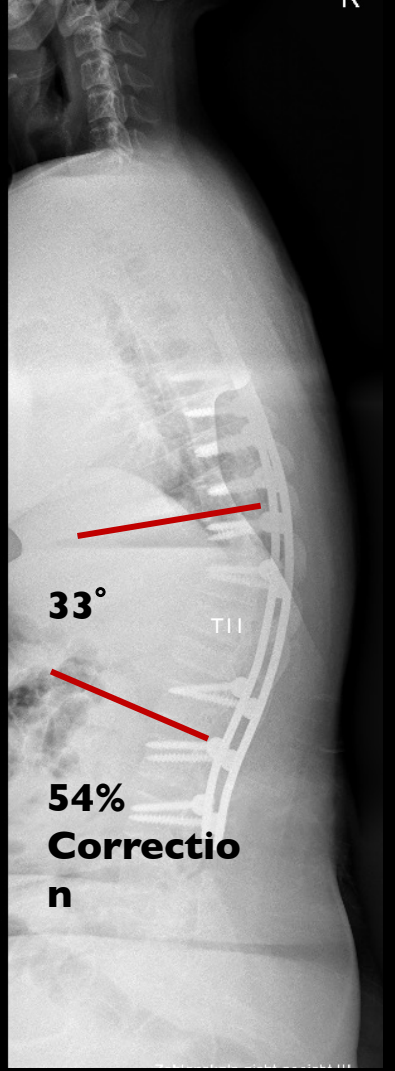
at 21 y/o:
mild paraparesis
after fall





Sx: multilevel laminectomies plus 2 level PSO plus fusion



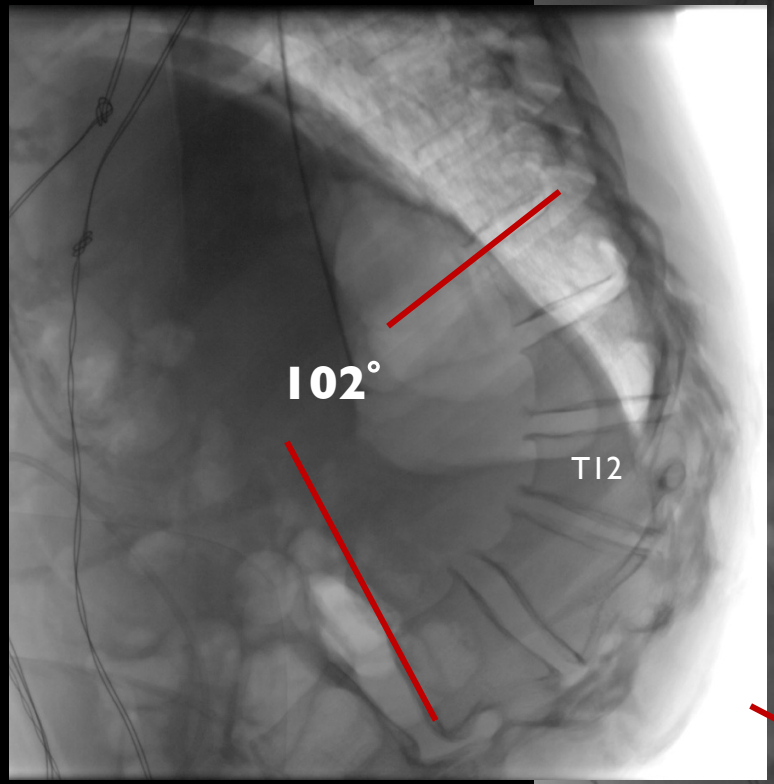


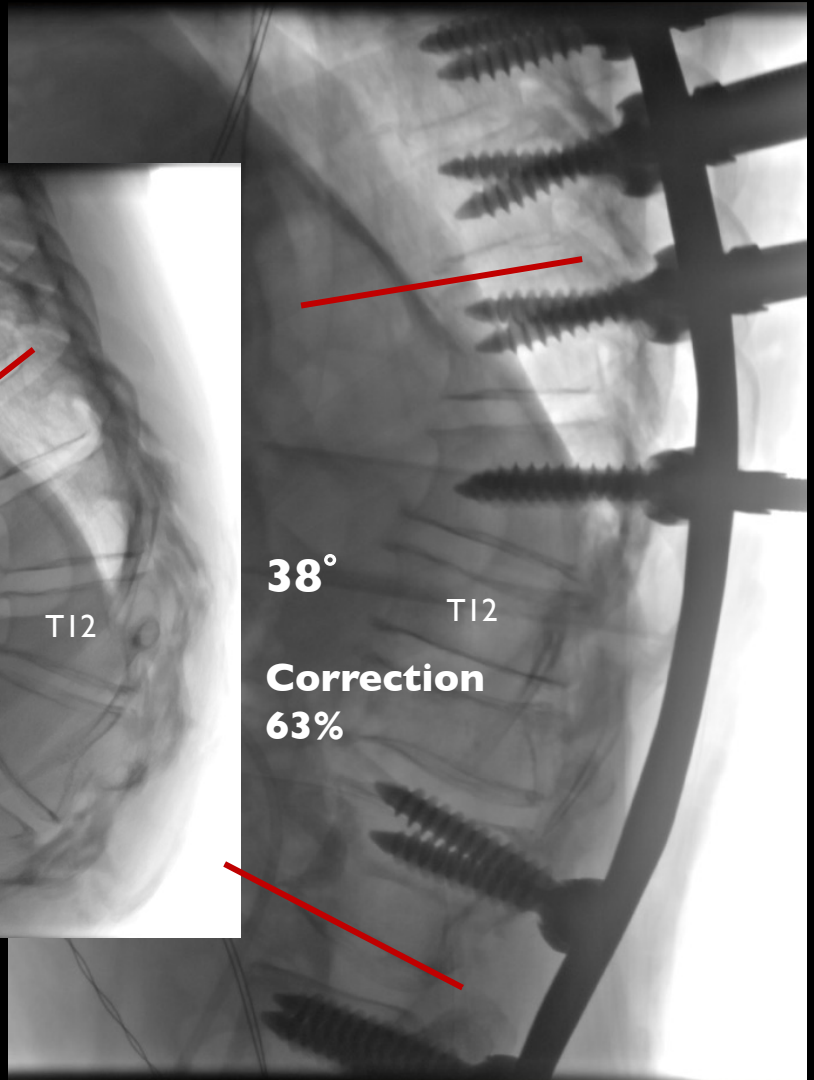
12 y/o boy,
routine MRI

at 21 y/o:
mild paraparesis
after fall

4 weeks post op
laminectomy T8-L2,
PSO T11 and T12

45y/o female
distinct paraparesis





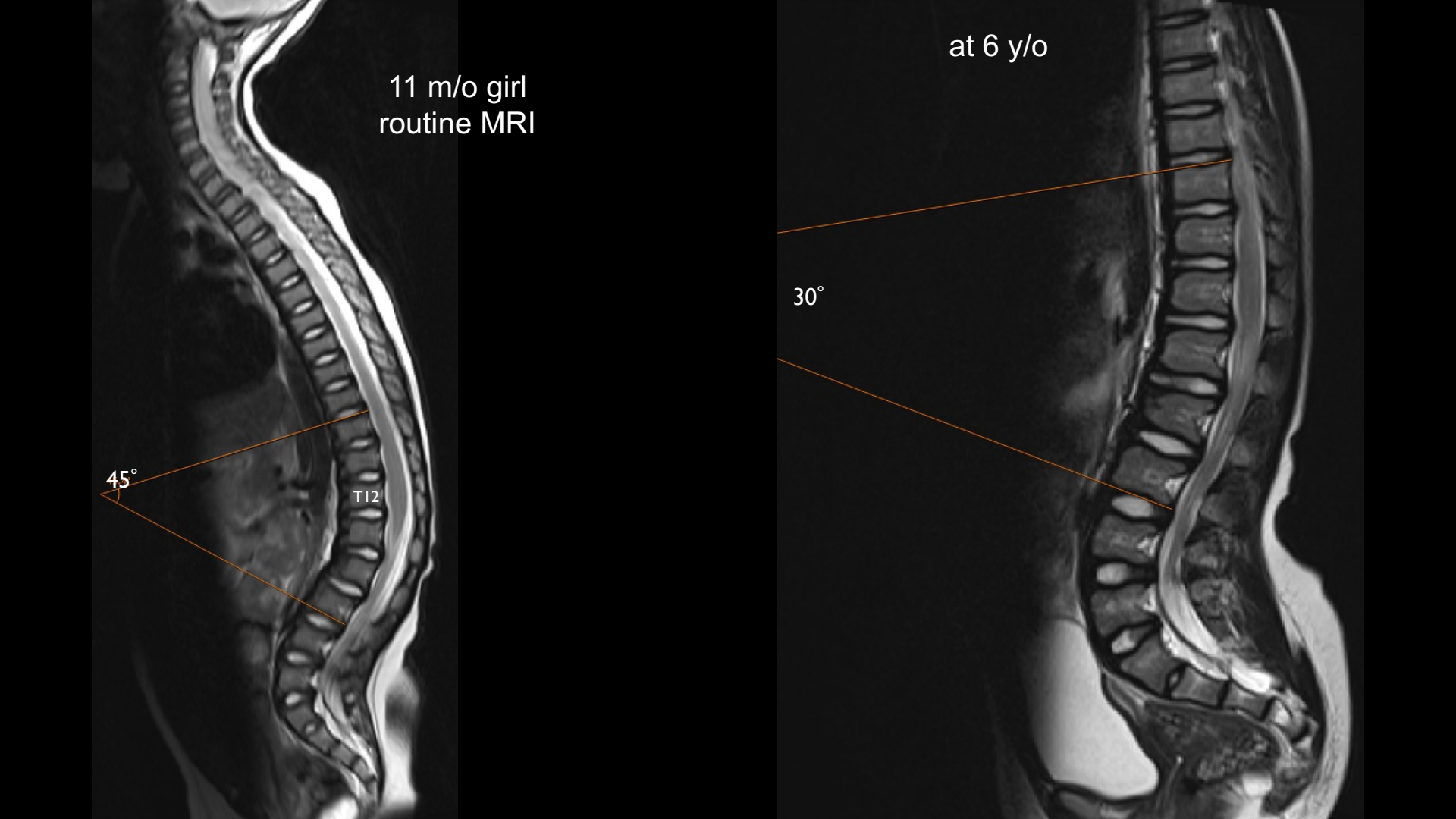

T12


T12

Medical Faculty Mannheim
Heidelberg University
University Medical Centre Mannheim

- 7 adults with ACHO (age 53 +/- 15 y.), 2008-17, Beijing
- Surgery: resection of 1-2 level, decompression of 5 +/- 2 level, fusion of 9 +/-2 level, correction: 73 +/- 15%
- Complications:
  - 14% dural tear
  - 71% neurological symptoms, 28% permanent
  - 57% surgical complications, 43% rod fracture, 14% PJK

Wang H. et al.: Posterior vertebral column resection (pVCR) for severe thoracolumbar kyphosis in patients with achondroplasia. Global Spine J. 2021


11 m/o girl
routine MRI
45°
T12
at 6 y/o
30°

Thoraco-lumbar kyphosis – natural history

- 60 children with ACHO (age at index presentation) 11 +/- 7 m., FU 5.7 +/- 3.6 y), 1997-2013, Johns Hopkins, Baltimore
- Initial curve: 44 +/- 11 degree
- At start of ambulation (21 +/- 8 months): curve <20° in 15%
- 1 year after independent ambulation : curve <20° in 58%
- Persistence of kyphosis at last FU:
  in 30% (Association with delayed motor developement)

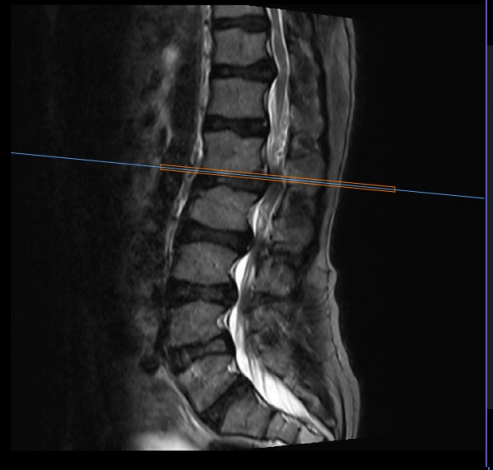
Margalit A. et al.: Walking out of the curve: Thoracolumbar Kyphosis in achondroplasia. JPO 2018

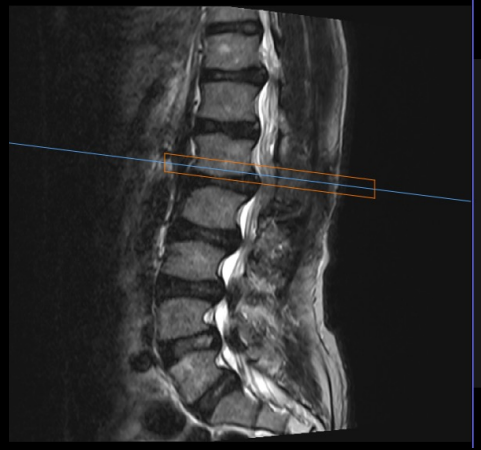
Medical Faculty Mannheim
Heidelberg University
University Medical Centre Mannheim

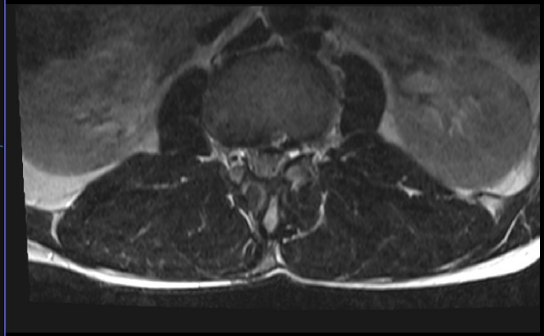
- First spine imaging: MRI at 6 month to 2 years in combination with Foramen magnum screening (nice to have)
- Clinical screening of kyphosis between 6 month and 4 years, at 4 years x-ray if kyphosis still clinical significant
- In patients with TLK >20° at age 4y -> regular (every 12 – 24 months) control of deformity progression via standing x-rays and/or clinical exam
- MRI thoraco-lumbar and lumbar if clinical signs (claudication, paresthesia etc.) of spinal stenosis occur

Goal: Timely initiation of surgery (lumbar decompressions / correction of kyphosis) before permanent neurological deficit is present

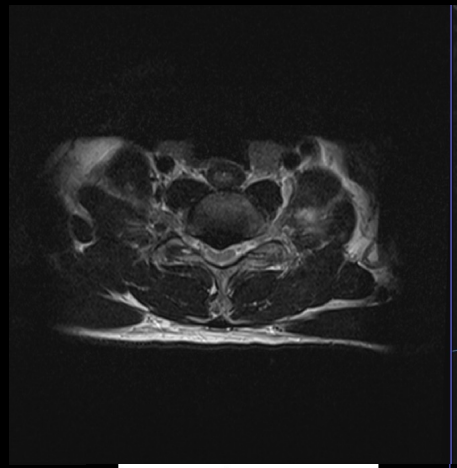
55y/o man, bilateral dropfoot for at least 5 years

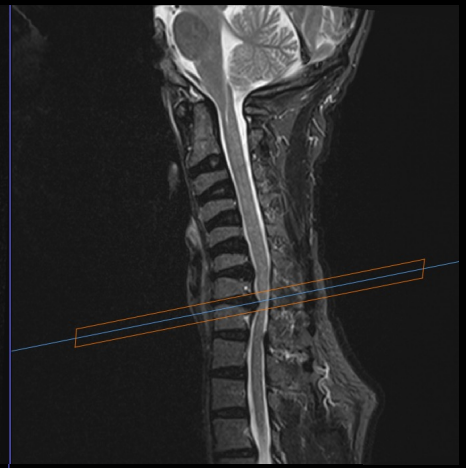






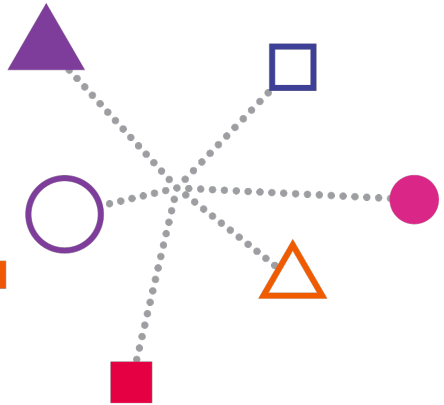
55y/o man, tingling paraesthesias right fingers for 3 months





Medical Faculty Mannheim
Heidelberg University
University Medical Centre Mannheim

- Neurological presentation of adult patients every x years to prevent late therapy initiation?
- Place for early bracing in children with delayed motor development for prevention of TLK?
- How is lumbar hyperlordosis and thoraco-lumbar hyperkyphosis interconnected?
- Need for more data! Register studies?



EAF

European Achondroplasia Forum