

# **Spine Pathology in Mucopolysaccharidoses**

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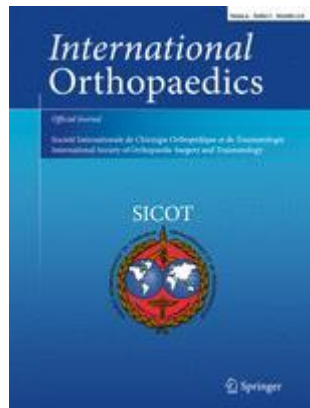
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# INTRODUCTION

- Mucopolysaccharidosis (MPS) are rare inherited metabolic diseases, causing lysosomal storage of muchopolysaccharides; clinical presentation involves skeletal system and particularly the spine.
- Anomalies include developing kyphosis at thoracolumbar junction, that can causes nervous symptoms, and dens hypoplasia with associated atlantoaxial subluxation that can cause myelopathy. We present our experience in the treatment of spine pathology in MPS.
- Medical treatments of MPS (hematopoietic stem cells transplantation, enzyme replacement therapy) seem to have little impact on spine disease: treatment of cervical instability often includes surgical decompression and stabilization, while thoracic lumbar kyphosis is treated by bracing and, in severe cases, with surgery. Bracing is more effective in kyphosis under 40° Cobb.
- Aims of spine surgery in MPS patients are restore vertebral alignment, protect nervous structures and stabilize the spine maintaining the best possible degree of mobility: our surgical cases in thoracic lumbar kyphosis treatment include the first ever described posterior approach only vertebrectomy in MPS and a case of lateral costo transverse approach instrumented correction

# MATERIALS AND METHOD

- From 2006 to 2018 we treated 22 MPS patients with spine diseases, mean age 6 years and 6 months
- All patients have been studied by spine plain radiographs and by MRI, integrated by CT scan in surgical patients.
- 19 patients with progressive thoracic lumbar kyphosis (mean 41° Cobb), without nervous symptoms, have been braced.
- 3 patients have been operated:
  - ✓ a 4 years 6 months old girl, quadriplegic, affected by spinal cord compression at occipitocervical junction, treated by decompression and occipitocervical stabilization
  - ✓ a 6 years old boy with progressive thoracic lumbar kyphosis, treated by posterior L2 vertebrectomy and instrumented arthrodesis T11-L4, with L1-L3 intersomatic cage;
  - ✓ a 7 years and 7 months old boy with progressive thoracic lumbar kyphosis with neurologic symptoms, treated by T11-T12 and T12-L1 discectomy by posterior-lateral approach, T11-T12 and T12-L1 intersomatic arthrodesis and T10-L4 posterior instrumented arthrodesis.



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**Spine challenges in mucopolysaccharidosis.**

[Crostelli M](#), [Mazza O](#), [Mariani M](#), [Mascello D](#), [Iorio C](#).

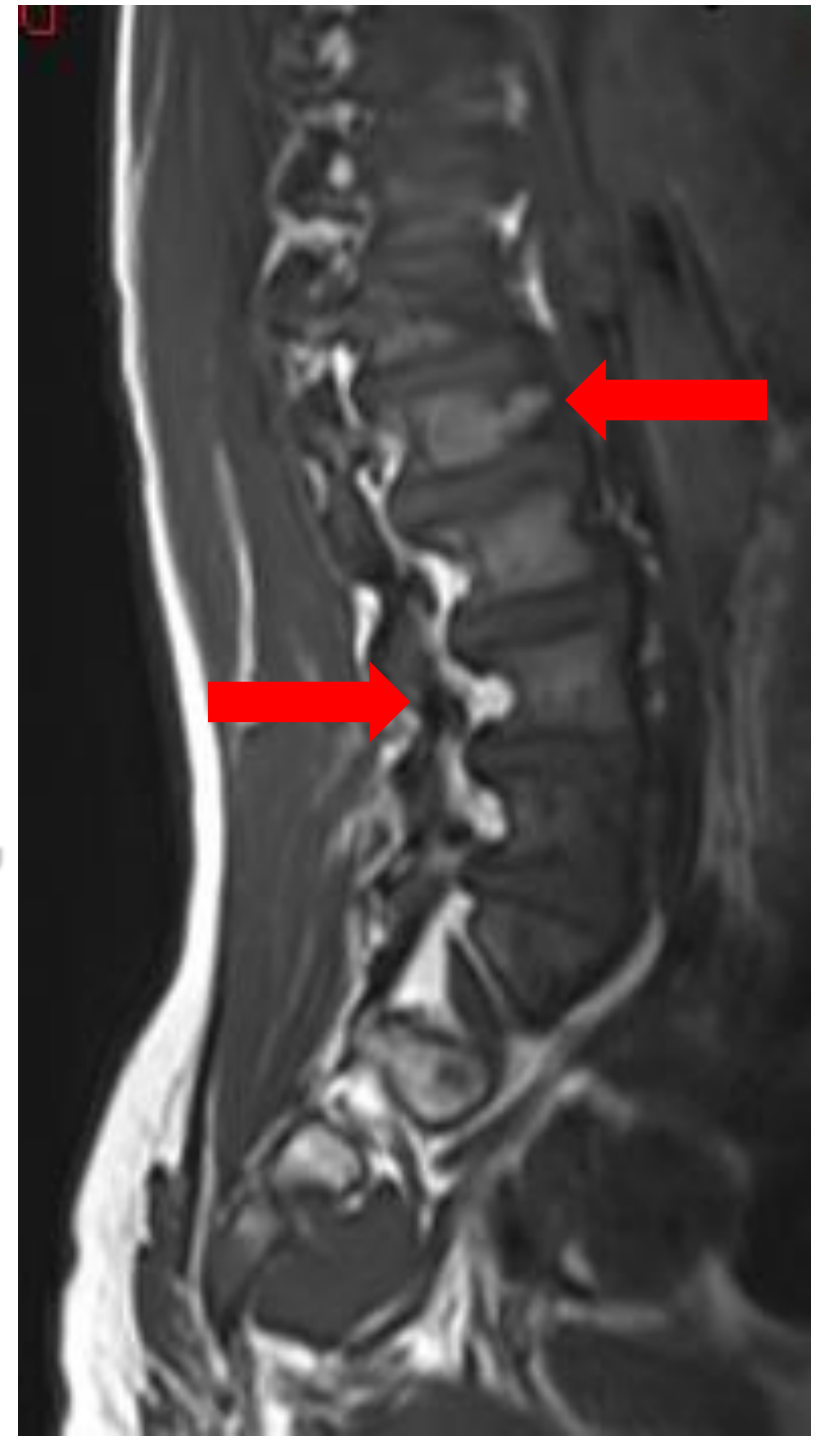
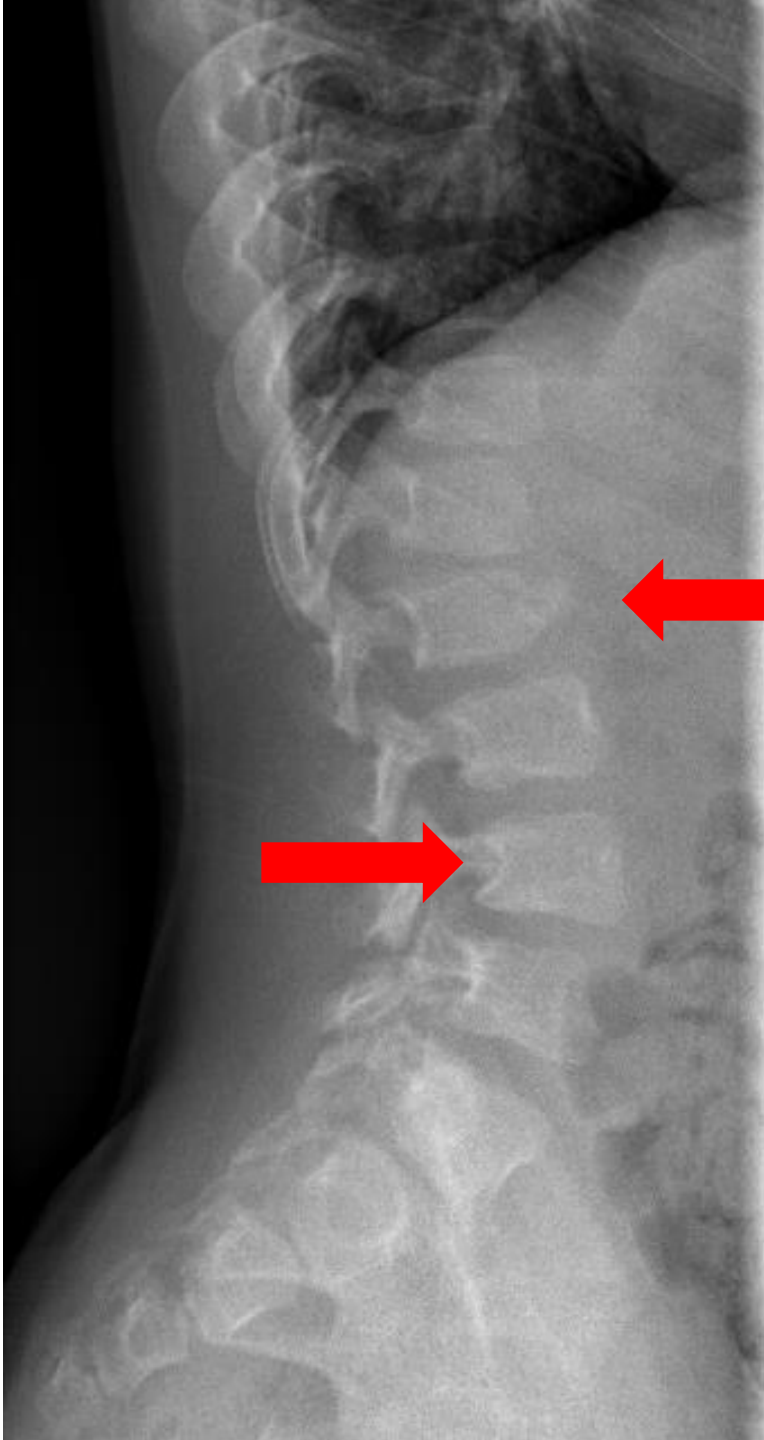
# Thoracolumbar kyphosis

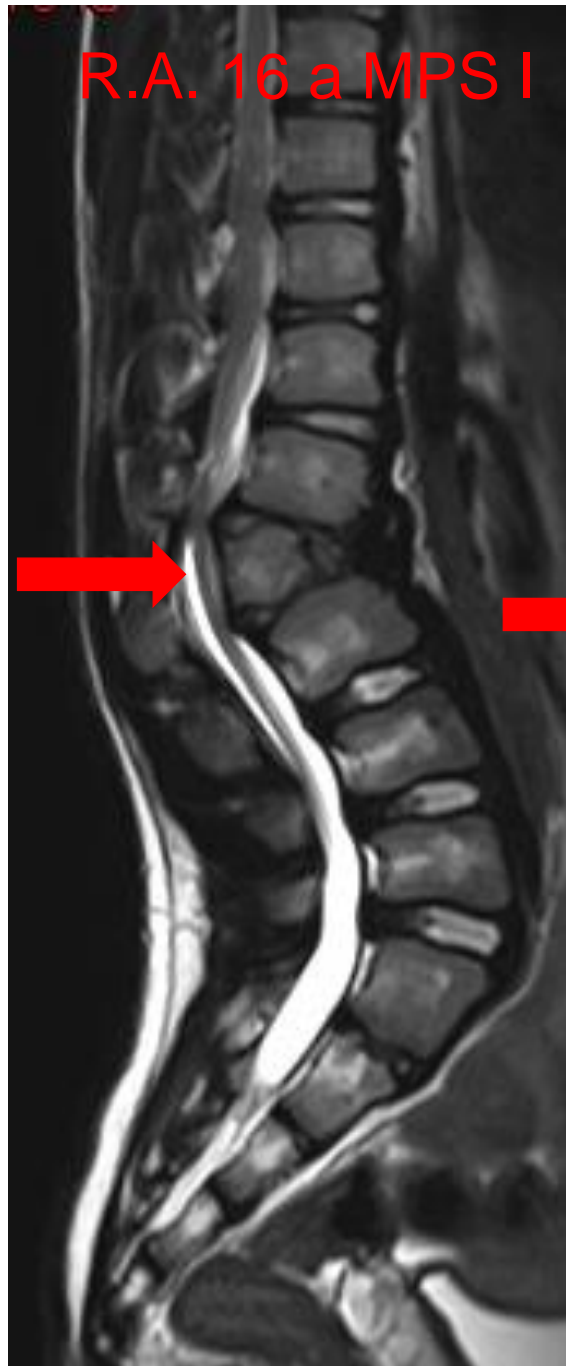
- ✓ Thoracolumbar (TL) kyphosis is a common spine deformity associated to Hurler syndrome (MPS I) and some authors consider thoracolumbar kyphosis almost universal in these patients once they are able to walk

**“BEAKED”  
VERTEBRA**

**“SCALLOPING”**

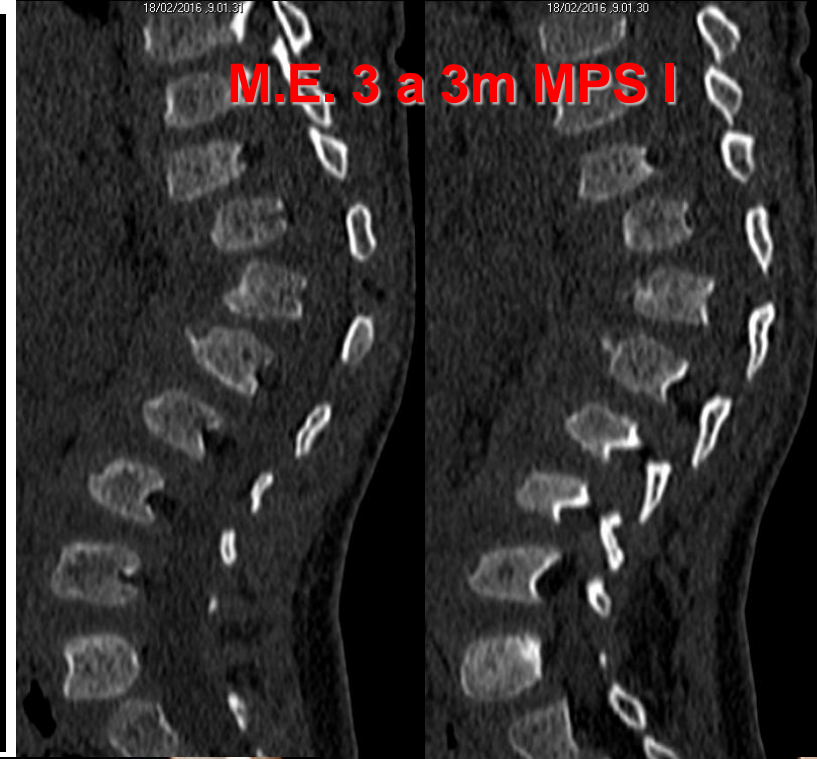
R.M. 7 a MPS I



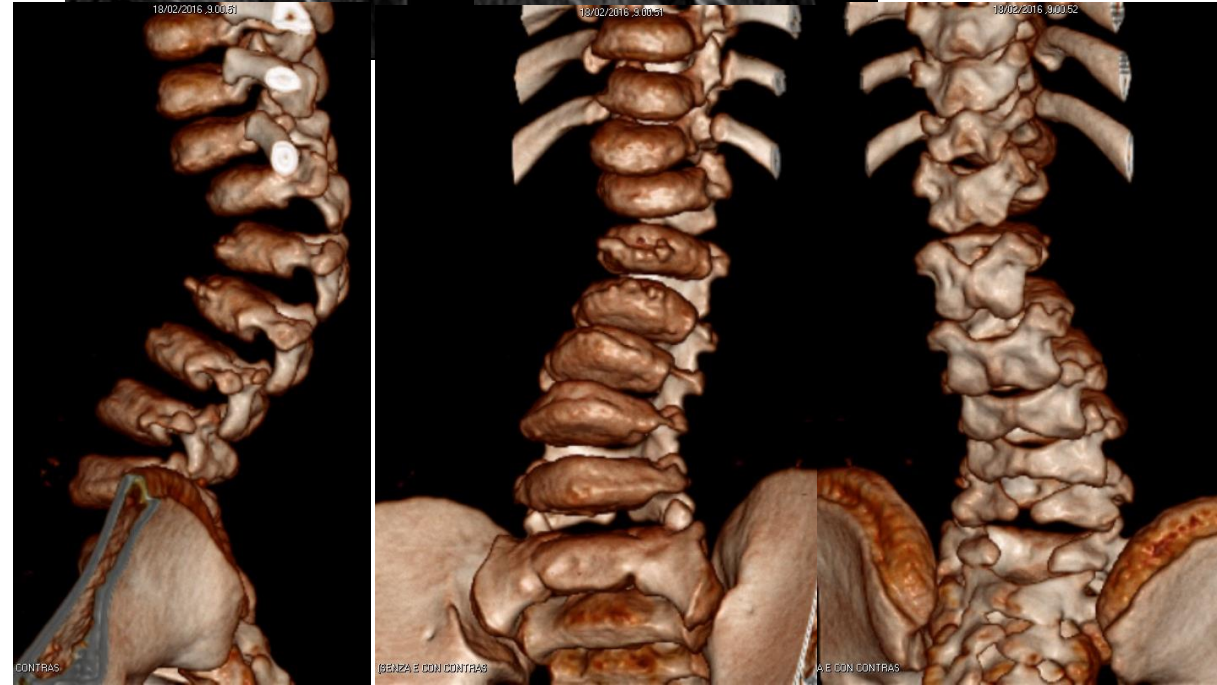


✓ The deformity does not improve with medical treatment due to lack of skeletal penetration of the replaced enzyme and does not always respond to orthosis treatment.





**M.E. 3 a 3m  
MPS I  
TAC 3D**



- ✓ Untreated kyphosis is likely to progress by posterior “listhesis” of “beaked” vertebra, finally causing nervous symptoms

# TL KYPHOSIS TREATMENT IN MPS: BRACING

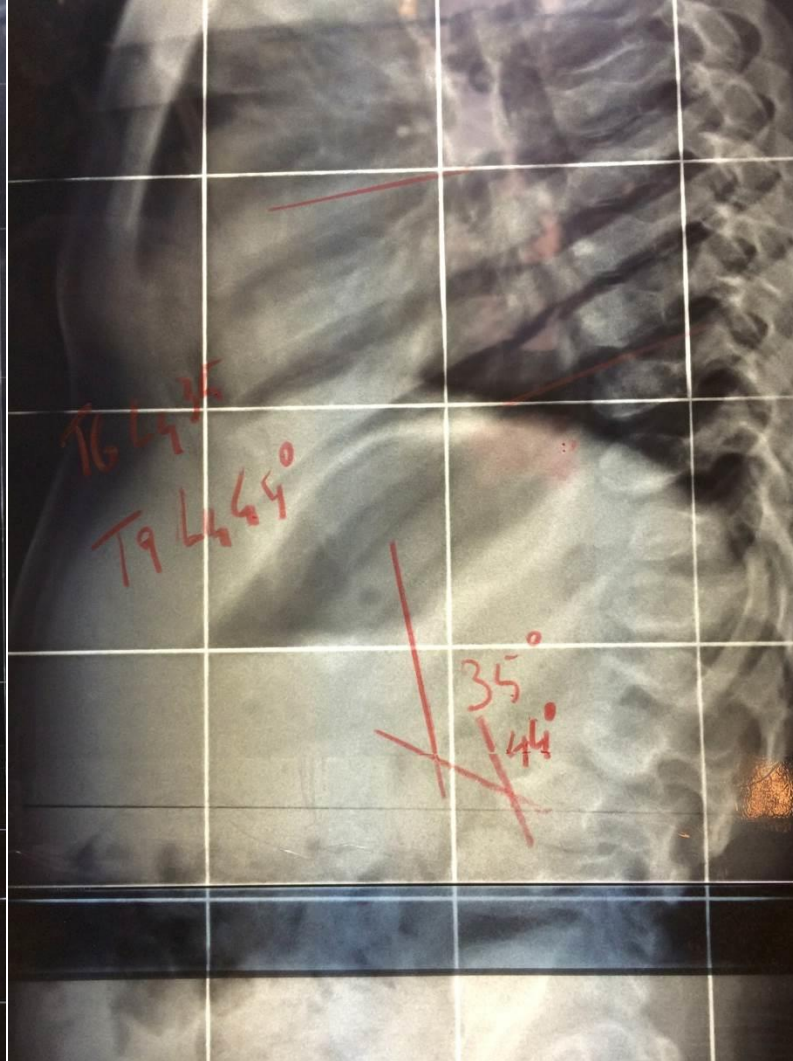
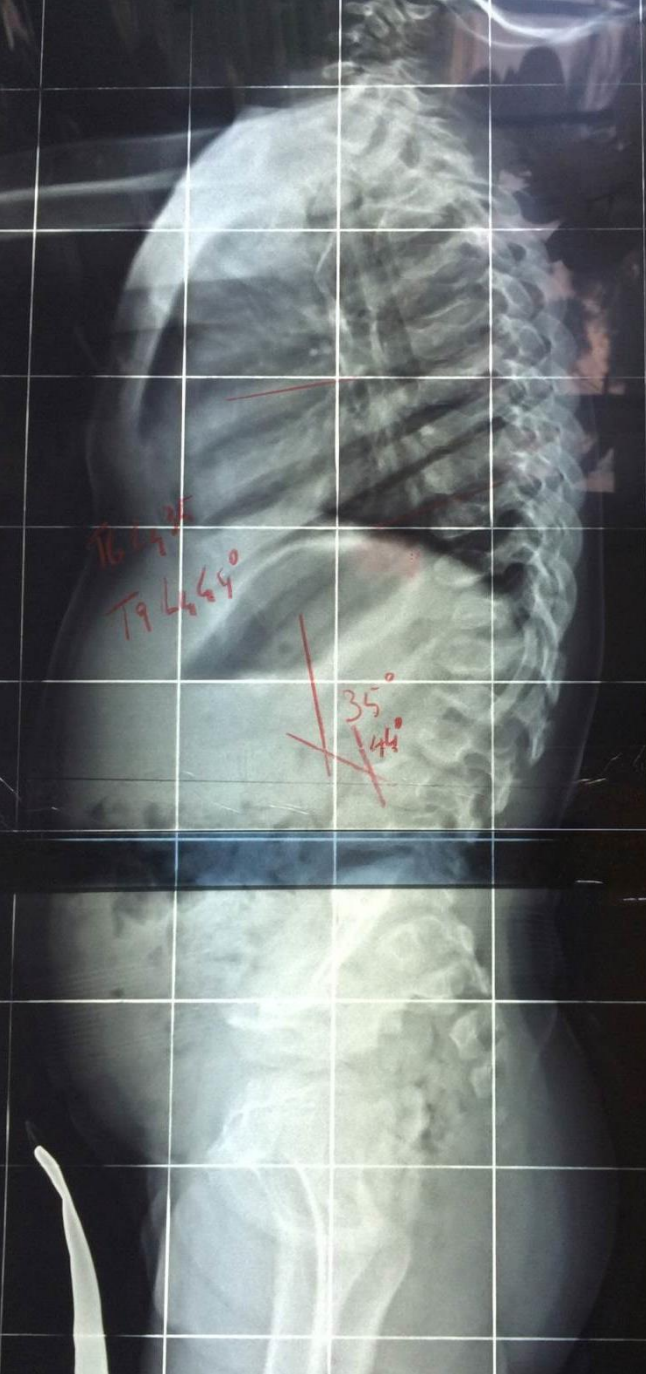
- ✓ We treated by bracing thoracic lumbar kyphosis in 19 patients with Hurler syndrome:
  - Mean age 6 years 3 months (range 4 years 9 months – 9 years)
  - Mean kyphosis at beginning of treatment 41° Cobb (range 36° -50° Cobb)
  - Mean follow up is 3 years 3 months (range 2 years 8 months – 5 years)
  - At final follow up mean Kyphosis was 25° Cobb (range 20° - 40° Cobb)
- ✓ Bracing should relieve “beaked” vertebra from weight forces but it is not sure to prevent deformity progression also in cases with good results at medium terms.
- ✓ Thoracolumbar kyphosis exceeding 40° Cobb has propensity to progress.





**C.A., 4 ys 9 ms,  
MPS I, 50° Cobb  
TL Kyphosis,  
treated by brace**



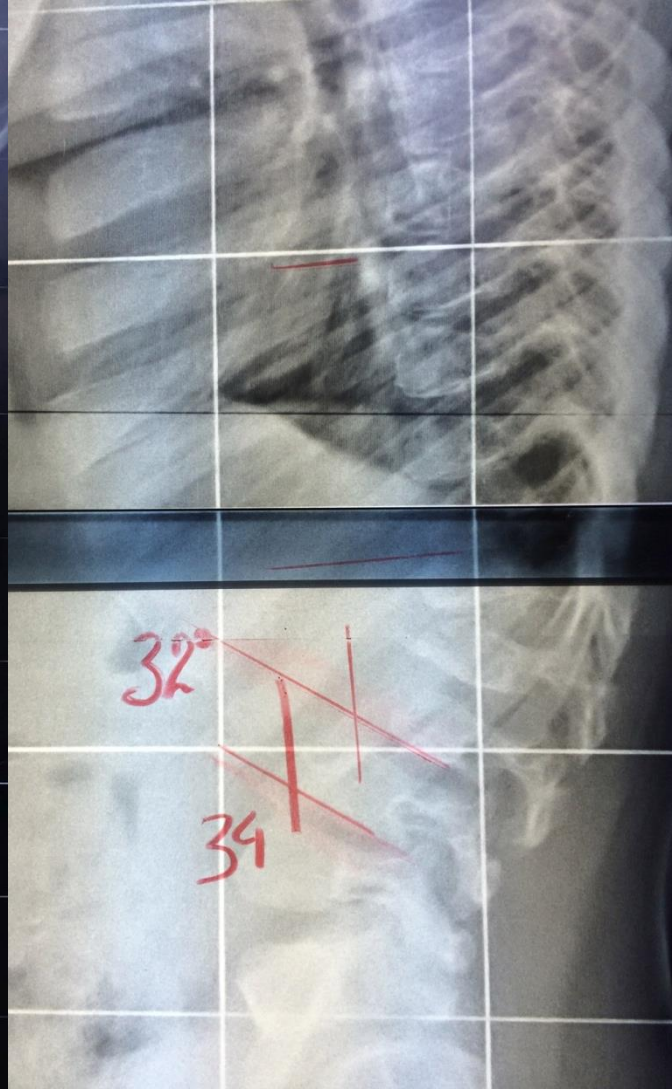


**C.A. 1 yr FU TL kyphosis reduced to 35° Cobb, segmental  
kyphosis reduced to 44° Cobb**



# GOOD CLINICAL RESULT

How will deformity evolve in time?  
Worsening of TL kyphosis during growth spur?



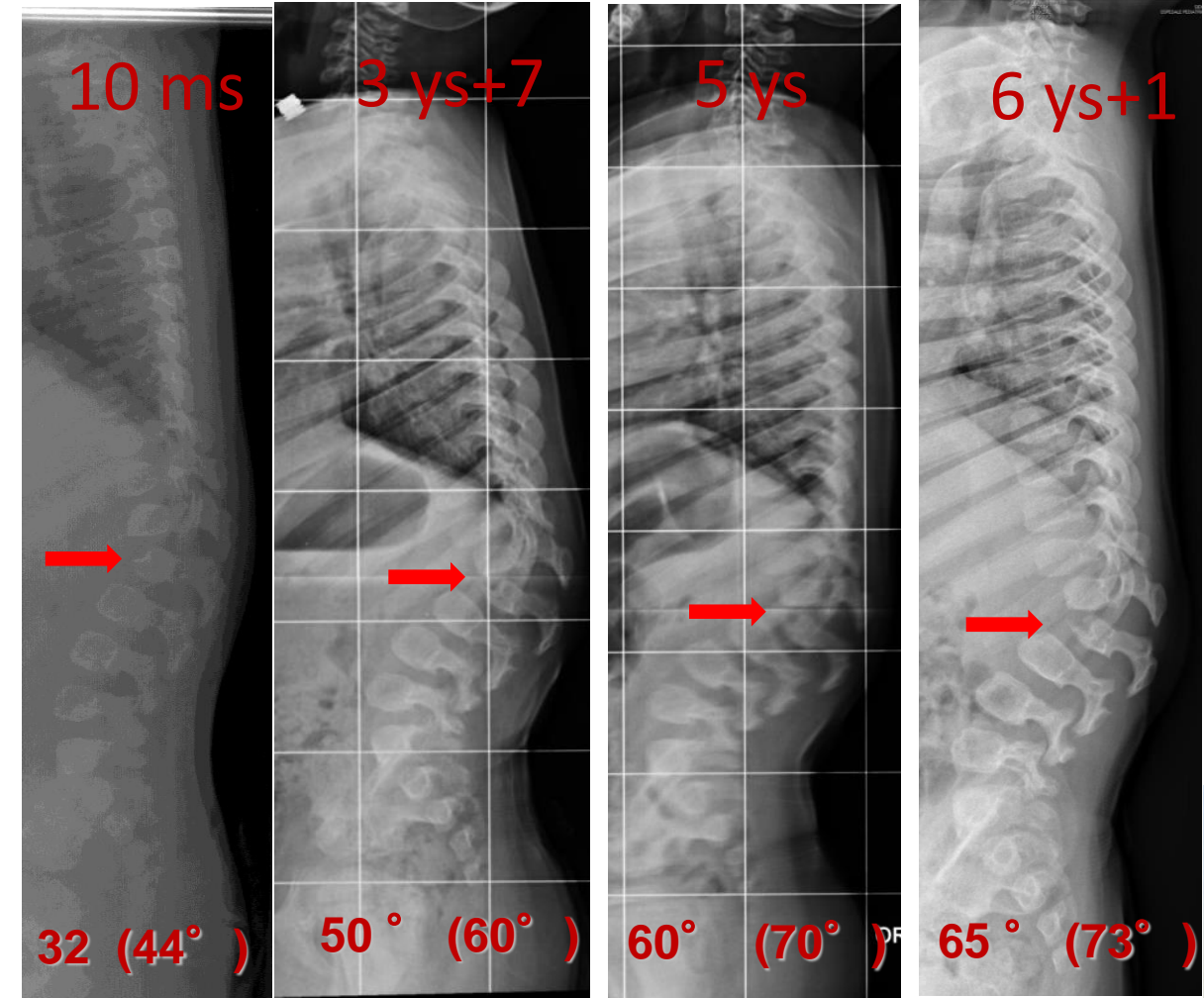
**C.A. 7 ys 5ms, 2 ys 8 ms FU, TL Kyphosis reduced to 32° Cobb, segmental kyphosis reduced to 34° Cobb**



# TL KYPHOSIS TREATMENT IN MPS: SURGERY

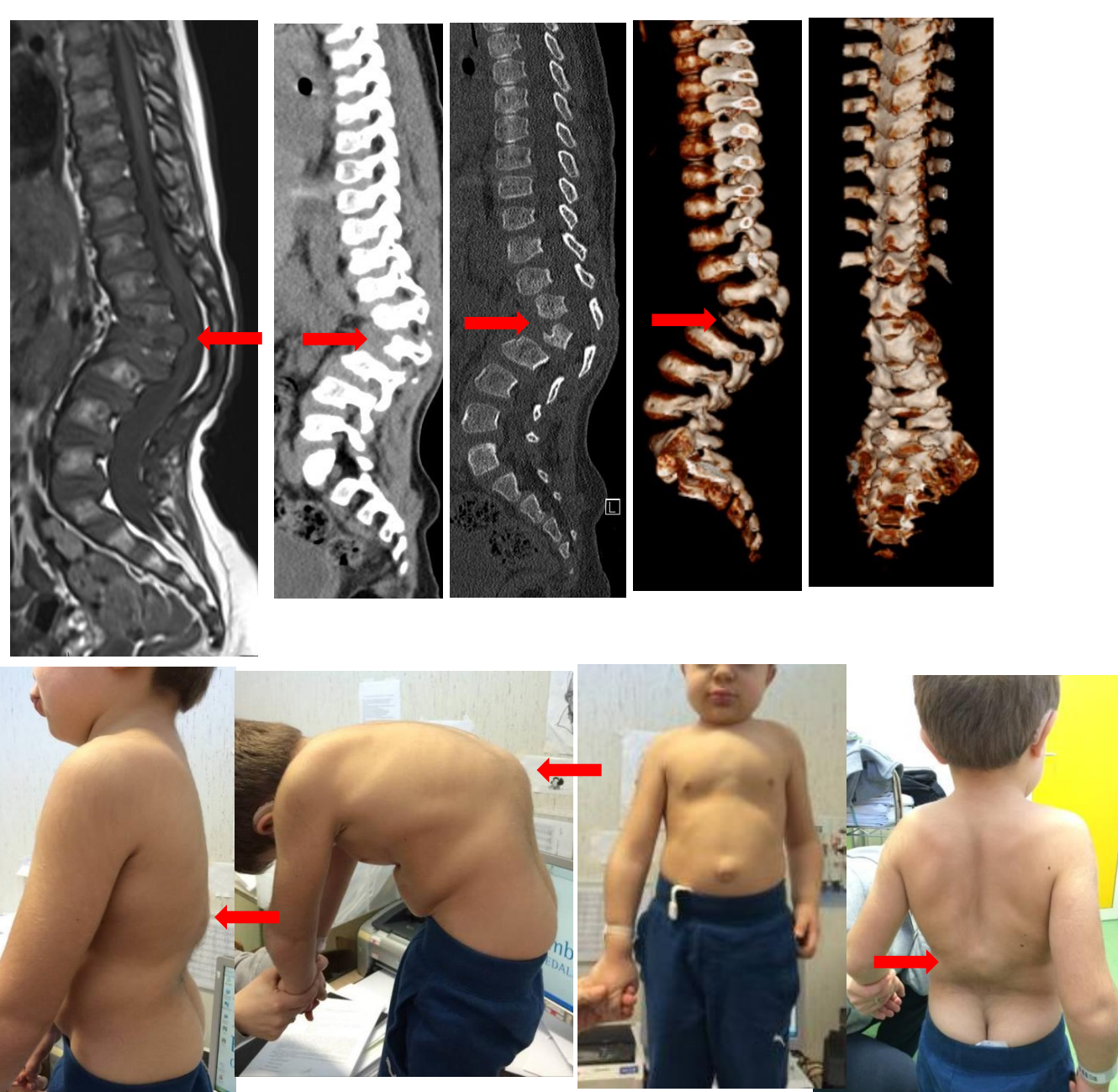
- ✓ When kyphosis is progressive and bracing is ineffective surgery is indicated to avoid neurological deficits, in analogy to the surgical treatment of progressive congenital kyphosis.
- ✓ Various surgical strategies have been proposed and, at present, there is no clear evidence of the best indication in the treatment of the deformity as for surgical technique and for intervention timing.
- ✓ We had no good results from previous experience with in situ fusion and we wanted to reduce the morbidity related to combined approach, anterior-posterior, as in treatment of congenital deformities.





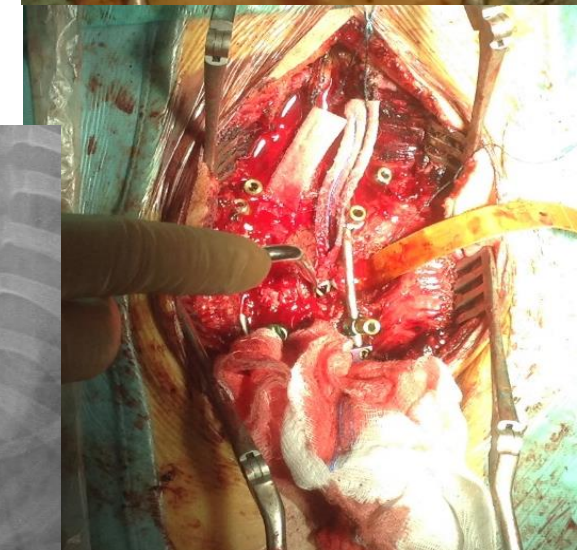
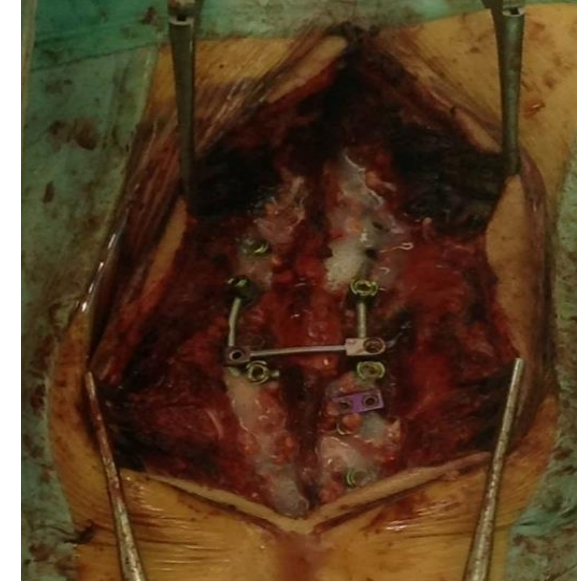
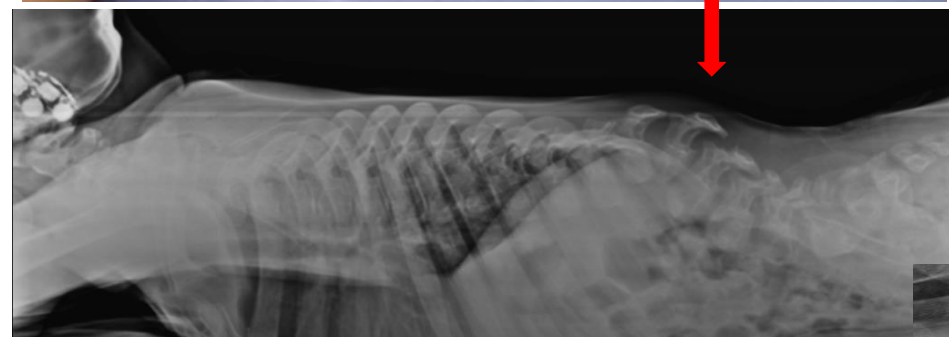
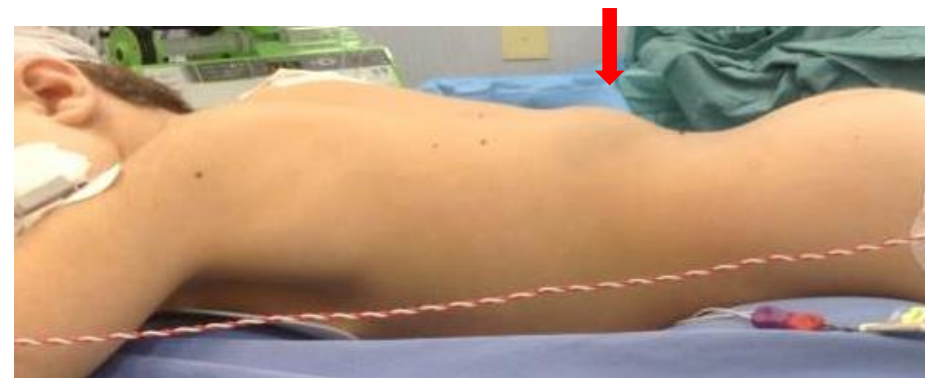
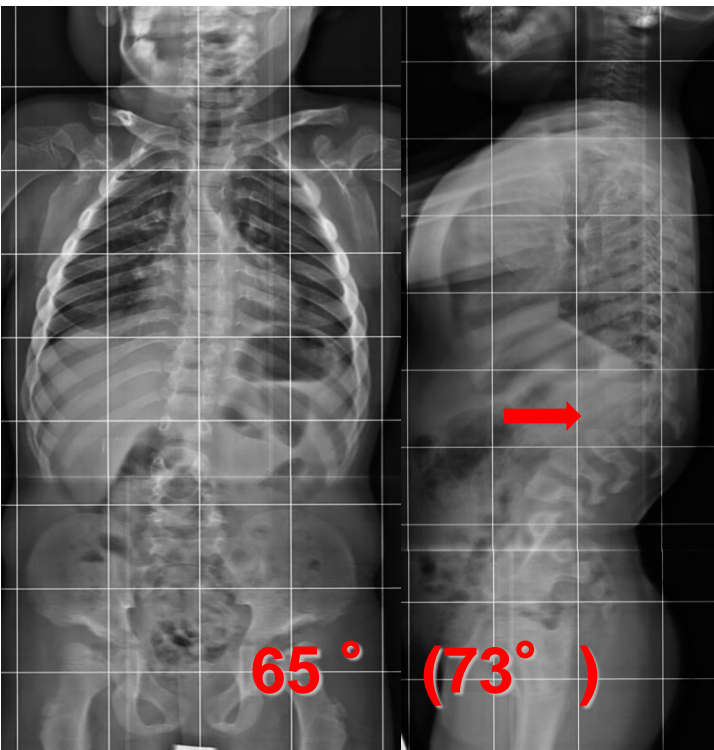
PROGRESSING TL KYPHOSIS

C.R., MPS I, treated by stem cells transplantation, progressing TL kyphosis, bracing has no result.

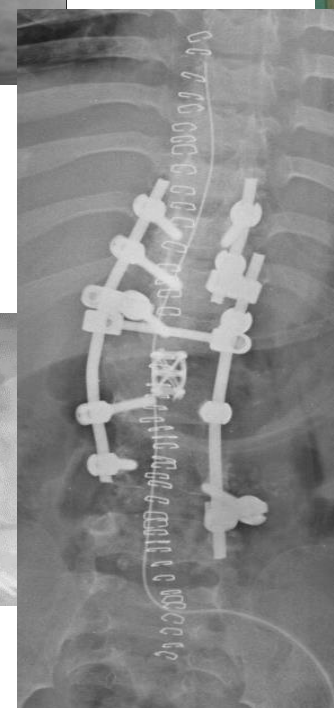
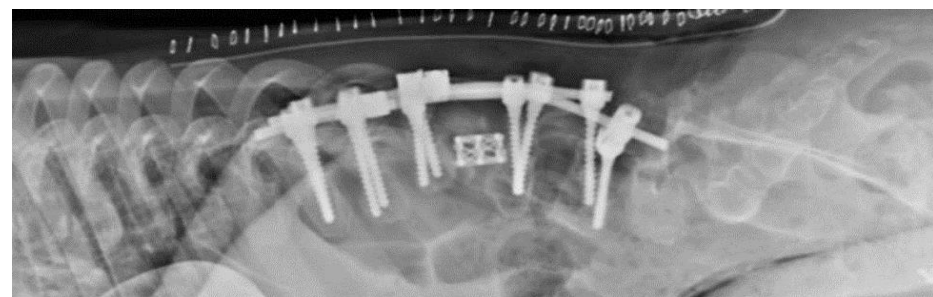


TORACIC LUMBAR HUMP





## STIFF TL KYPHOSIS



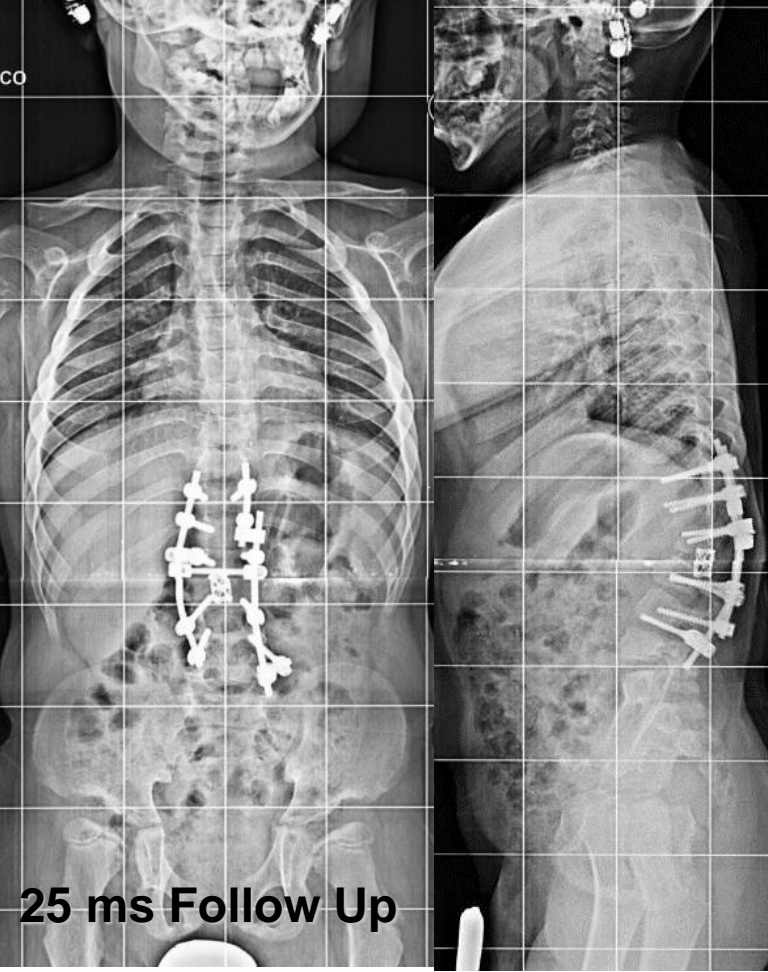
22° (37°)

✓ Posterior L2 vertebrectomy and instrumented arthrodesis T11-L4, with L1-L3 intersomatic cage.

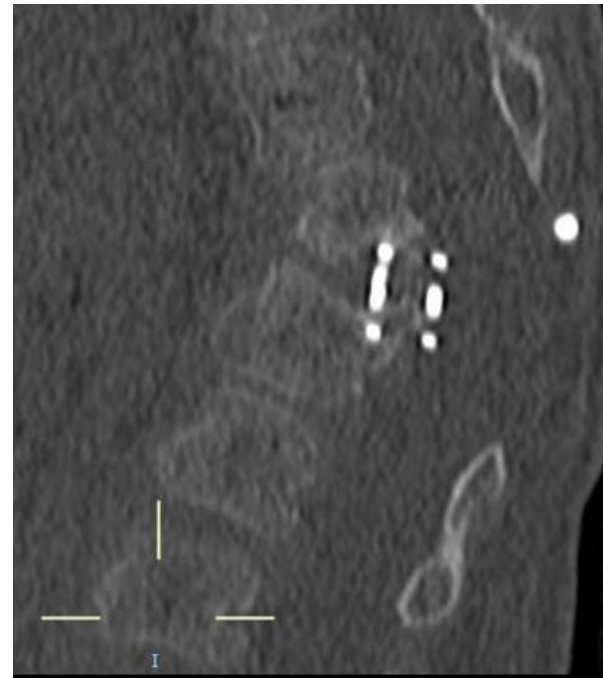
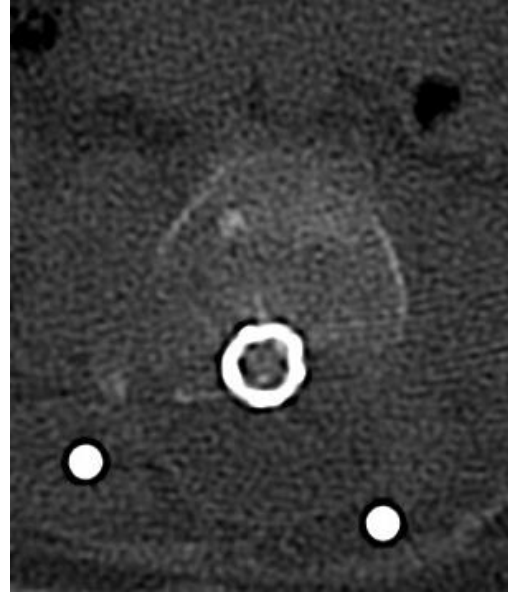
✓ To our knowledge, our patient is **the first case of thoracolumbar kyphosis in MPS 1 treated by posterior only approach total vertebrectomy.**

✓ We used the smaller adult cervical cage available, 1 cm in diameter.

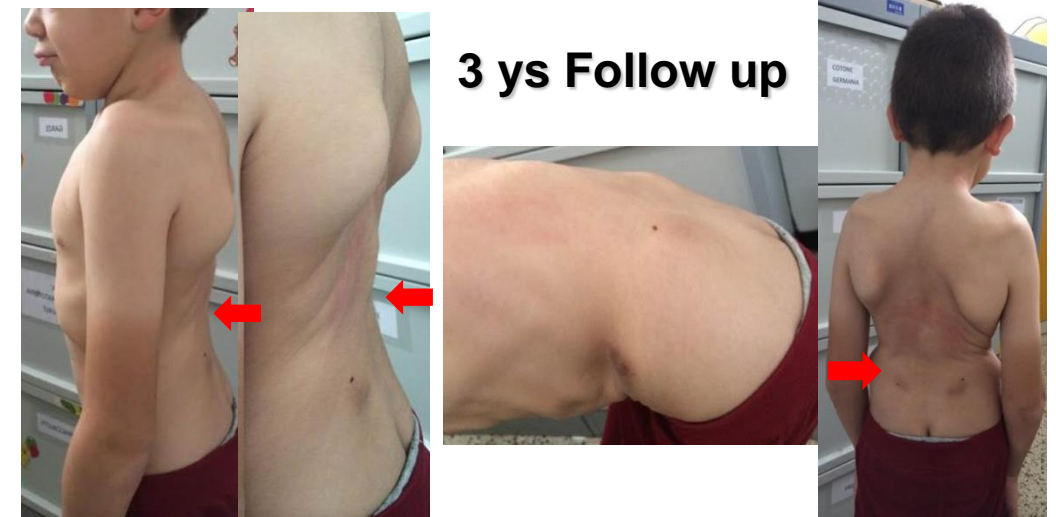
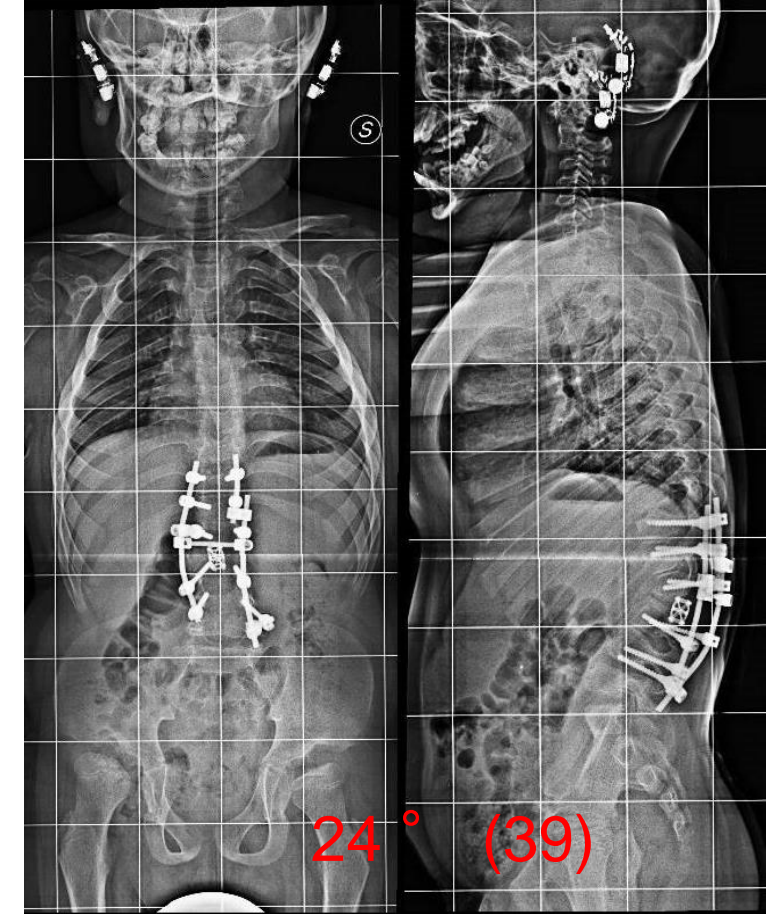




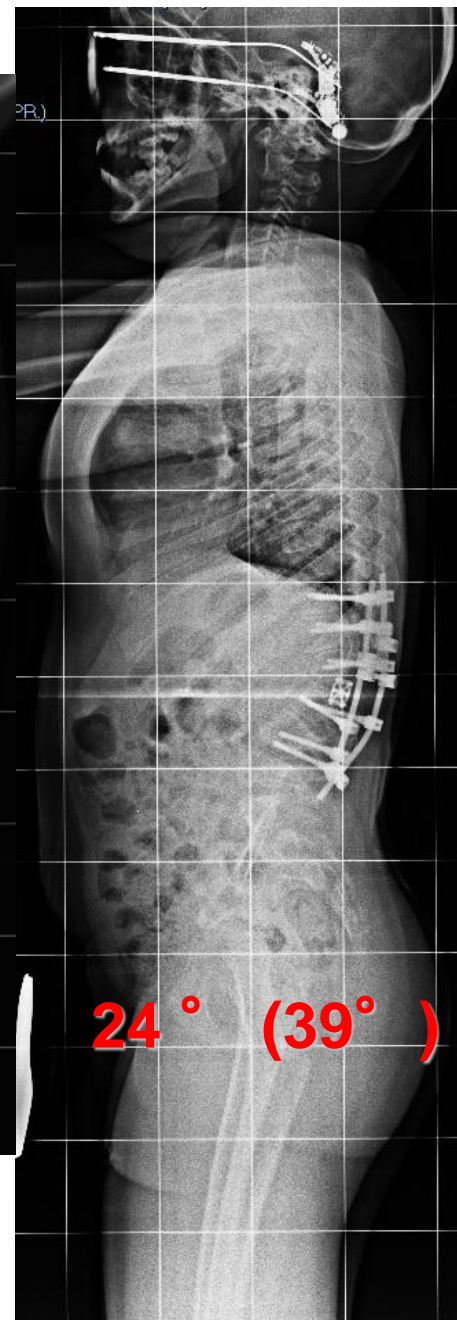
- ✓ Sub-optimal cage positioning, as limited vertebrae dimensions (vertebral plate diameter was less than 1cm) did not allow an adequate positioning even of the most minimal device available, resulting in an oblique position of the cage.
- ✓ Still the bone filled cage allowed a complete interbody fusion.
- ✓ The incomplete kyphosis resolution, with good cosmetic results and stability in the time, in our experience could reduce and avoid the occurrence of proximal junctional kyphosis.



**CT 27 ms Follow Up**





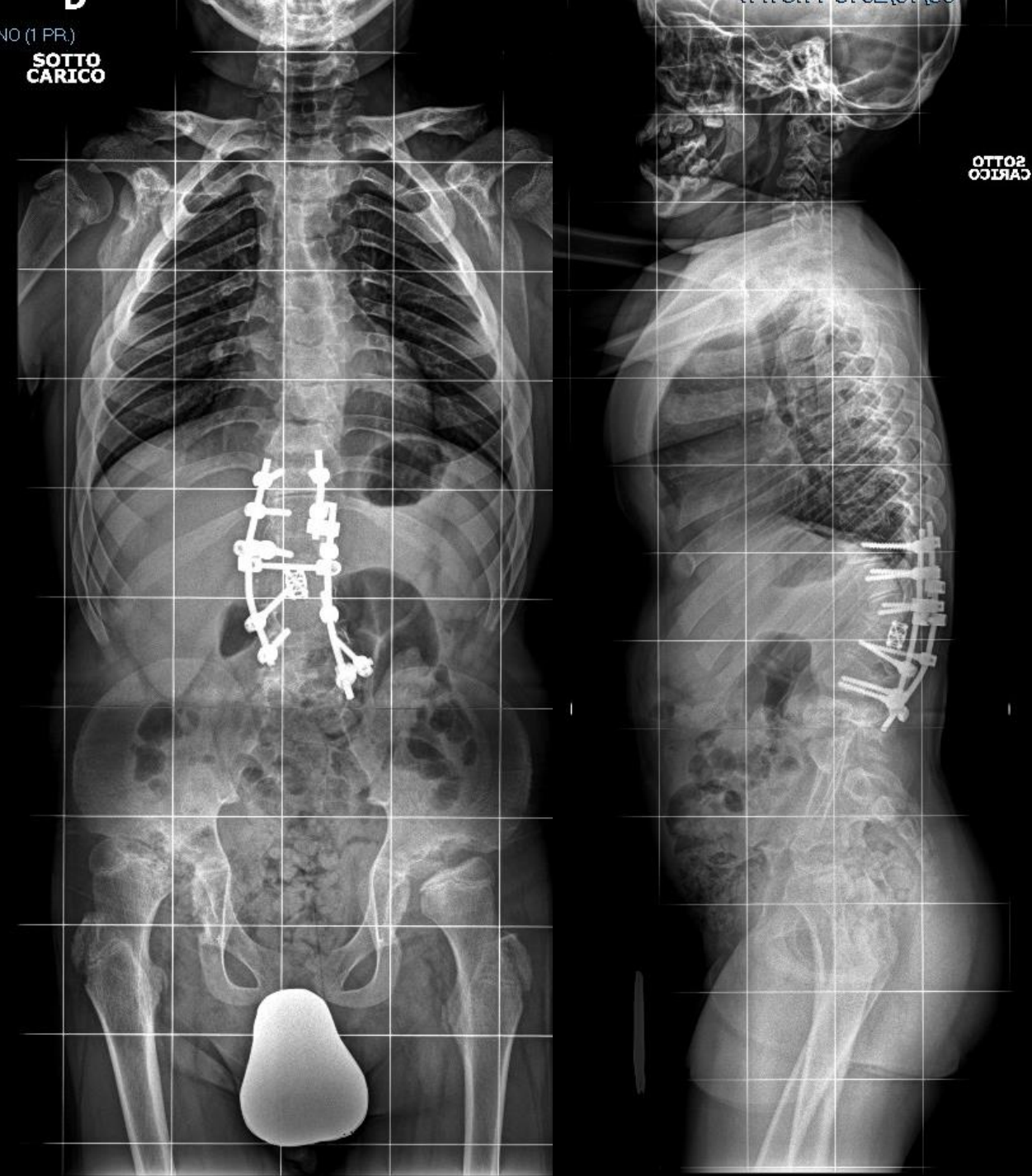


65 ° (73° )

24 ° (39° )

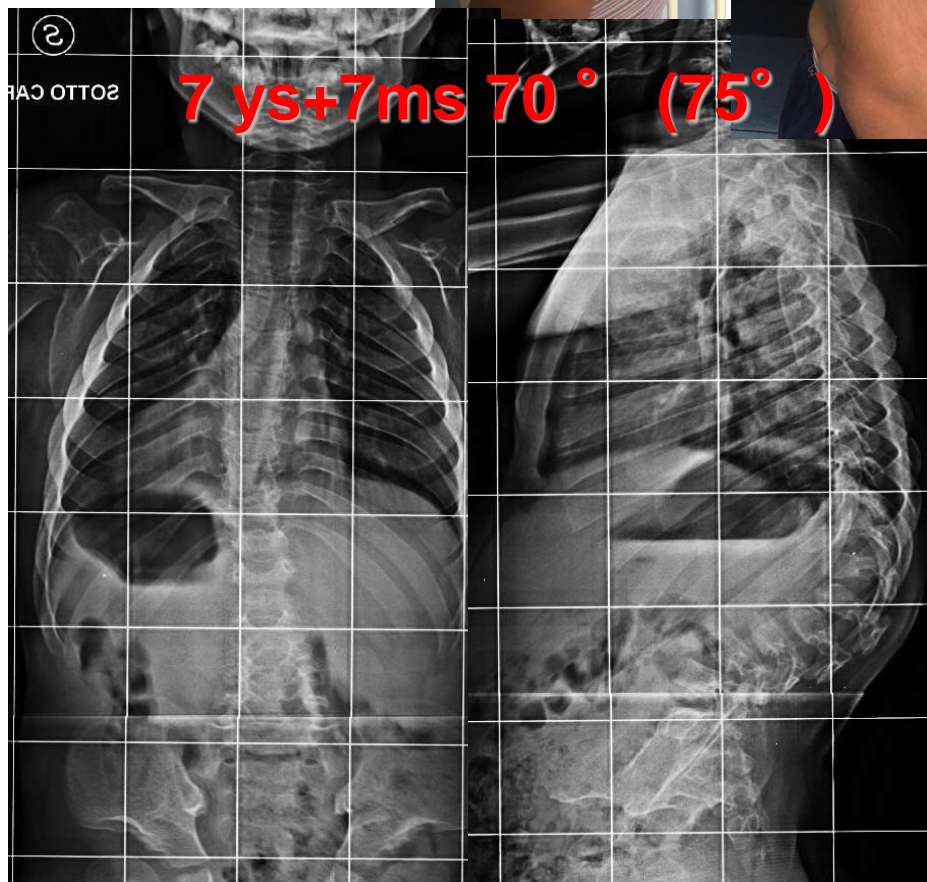
C.R. 10 ys at 4  
ys FU





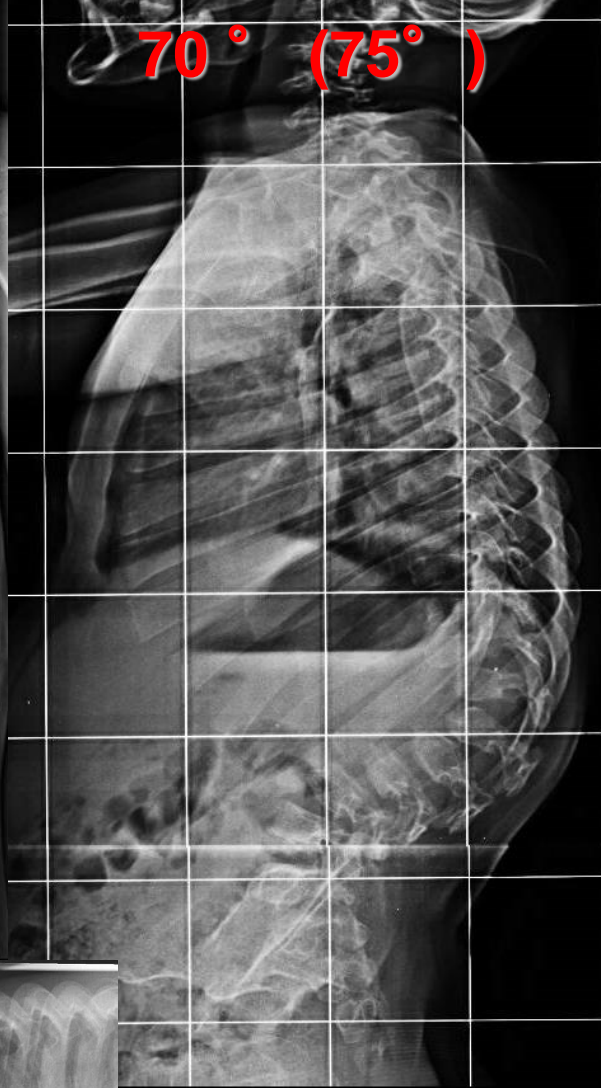
C.R. 5 ys FU, Genu valgus treated by temporary proximal tibia bilateral hemiepiphysiodesisi





- ✓ M.E. 7ys 7 ms, MPS I, progressive TL kyphosis, unsuccessfully treated by brace.
- Nervous symptoms:
- paresthesia in both legs
  - limbs somatosensory evoked potentials slowed down.

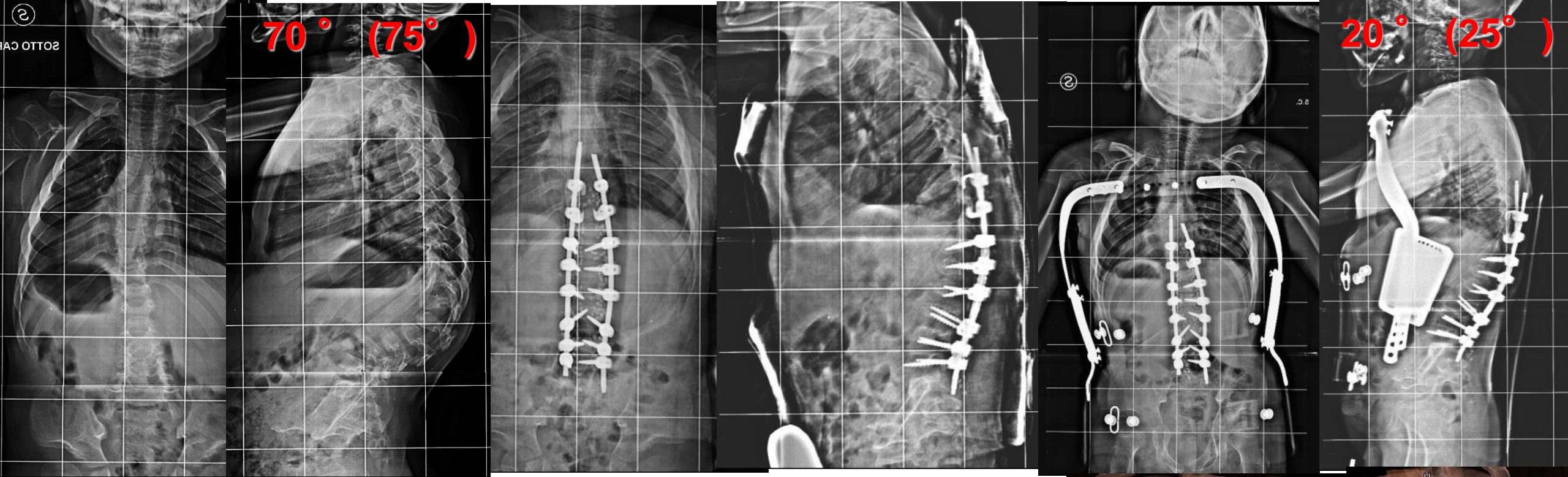




**TL Kyphosis is partially reducible in extension**

- Different surgical approach from the first case, to avoid problems of cage positioning and to avoid blood loss from vertebral osteotomy.
- Right lateral costal transverse approach, T11-T12 and T12-L1 discectomy, T11-T12 and T12-L1 interbody fusion with autologous bone graft from rib and posterior instrumented arthrodesis T10-L4.

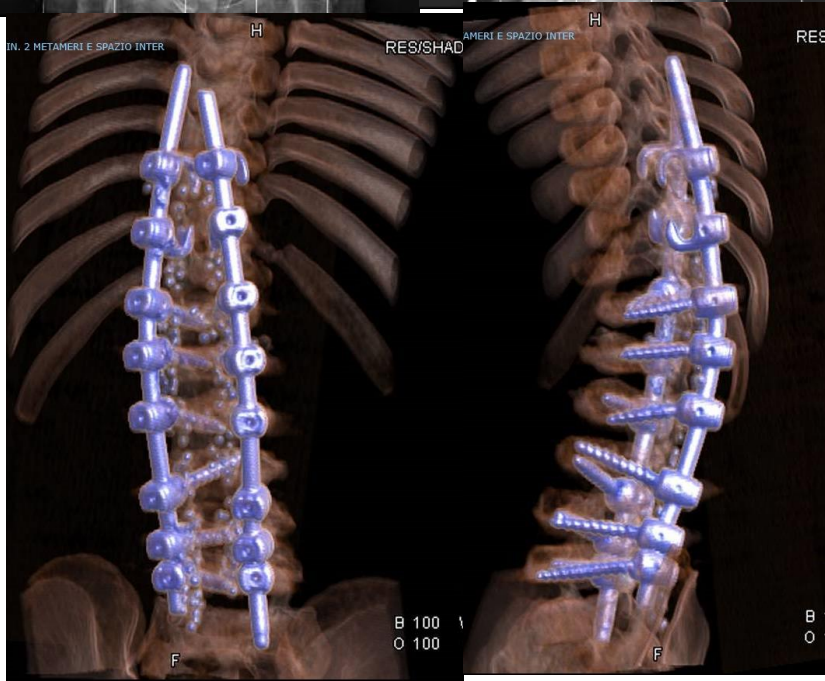




M.E. braced by cast for 8 weeks and then by brace for other three months.

5 months Follow Up CT scan exam shows complete posterior and lateral arthrodesis and limbs evoked somatosensory potentials show normal values.

In this patient too, we choose an incomplete kyphosis resolution, but with good cosmetic and functional results, to avoid the occurrence of proximal junctional kyphosis.







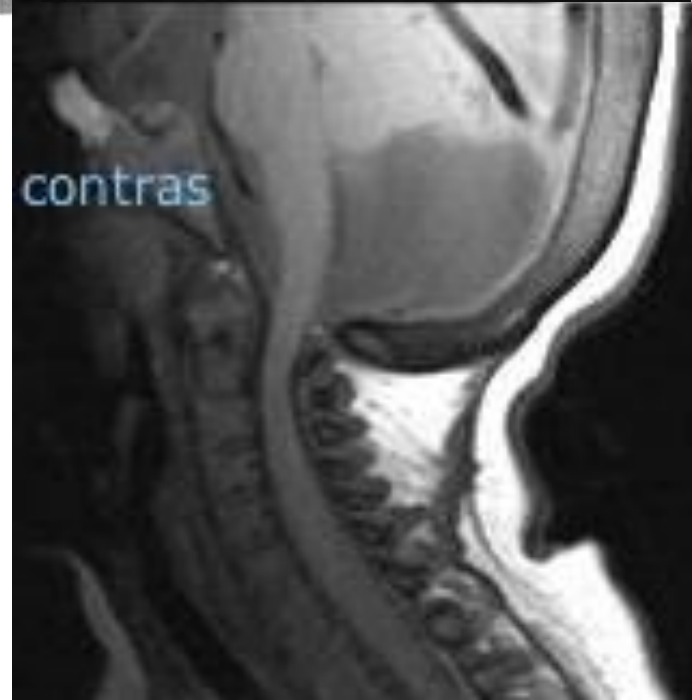
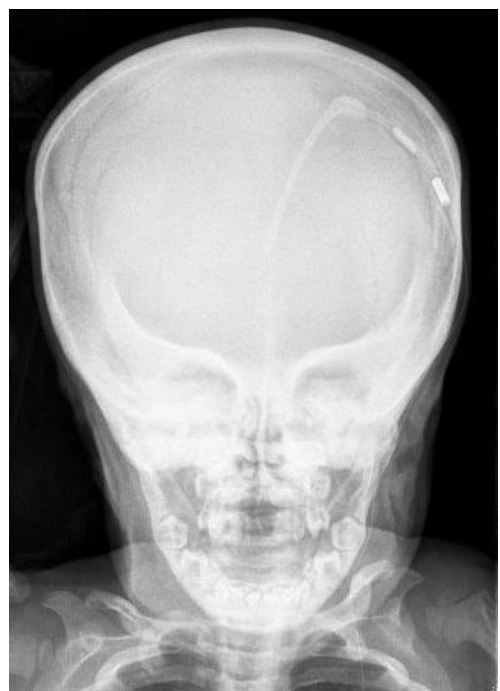
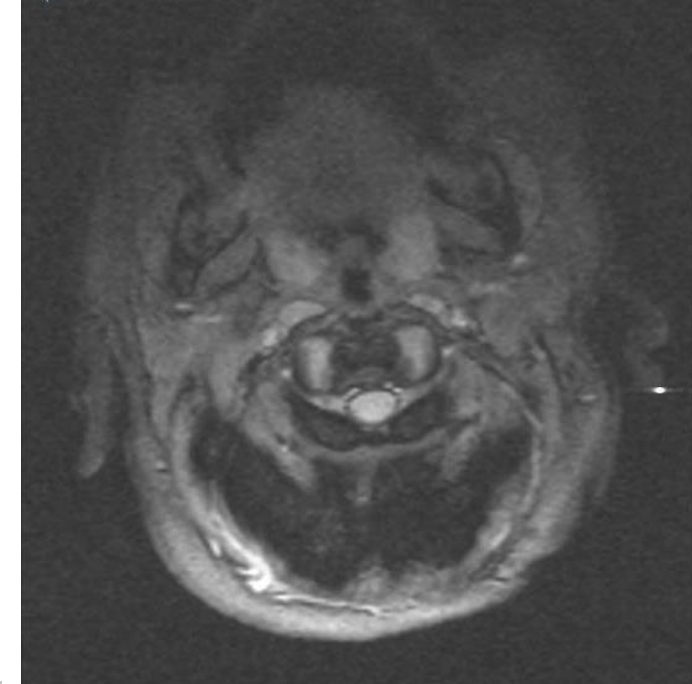
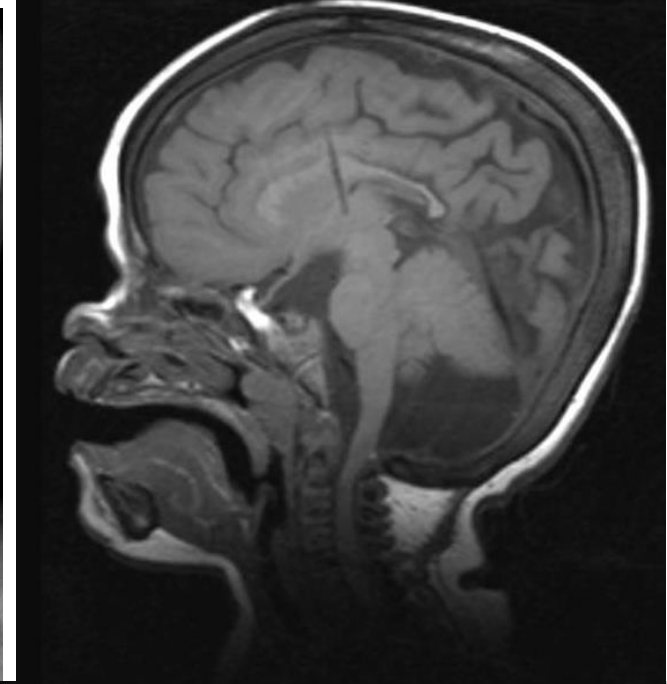
**1 year Follow Up**



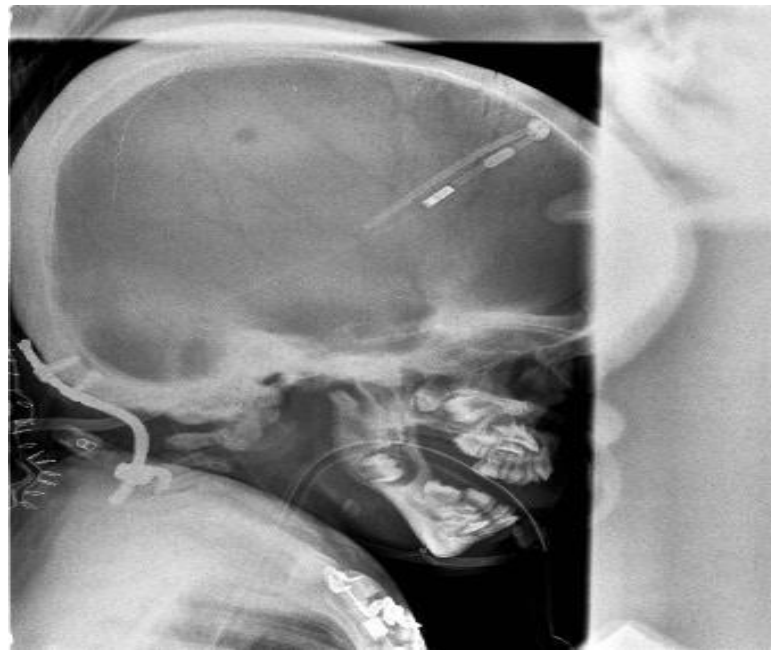


# MPS atlantoaxial instability

- In MPS atlantoaxial instability and spinal cord compression caused by GAGs deposits in peri-odontoid tissues is common and some authors recommend prophylactic fusion, eventually associated with cord decompression, at an early stage of disease in these patients
- Posterior occipitocervical fusion is the most common procedure in treating MPS occipitocervical junction instability, but patients with severe spinal cord compression may require decompression and posterior occipital cervical fusion

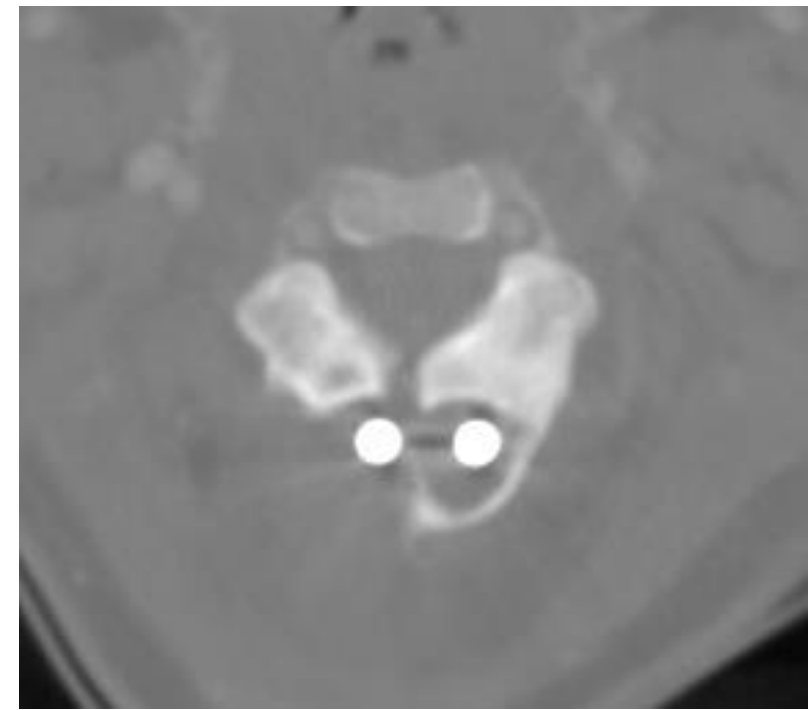


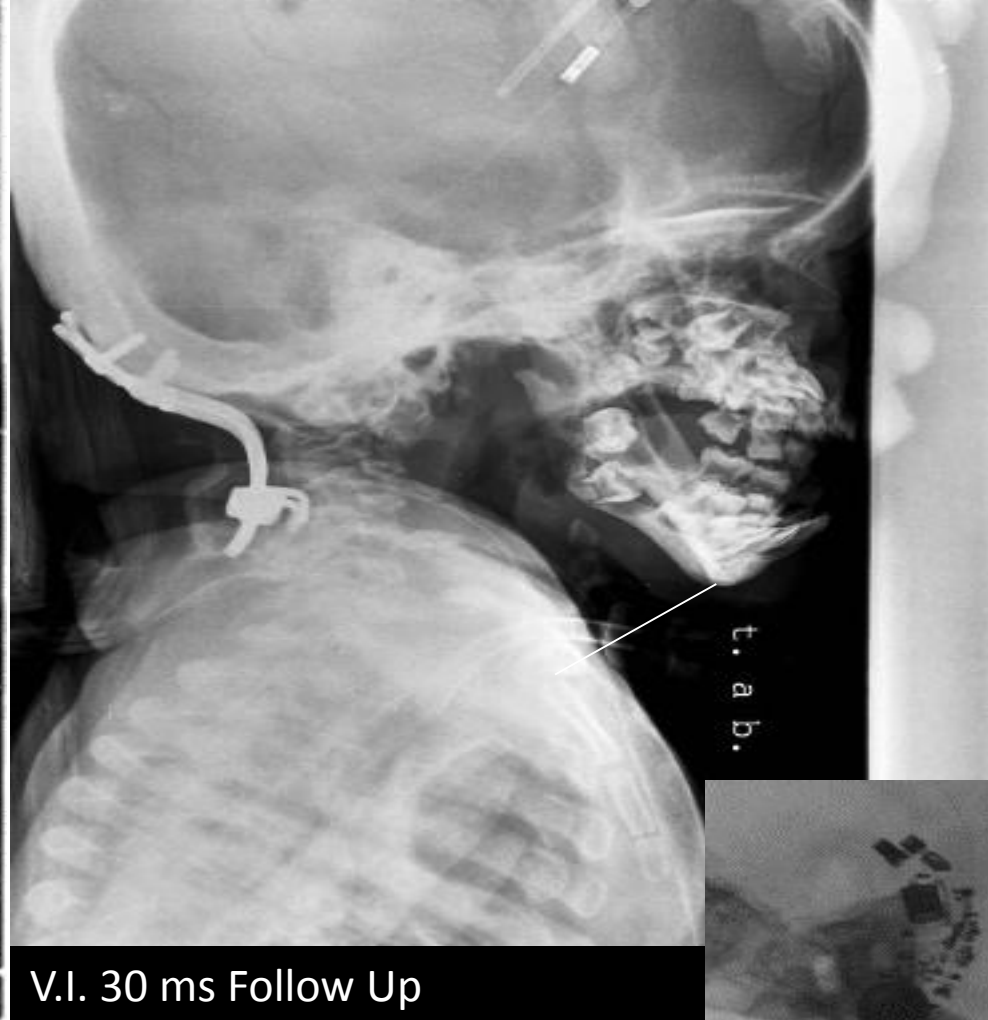
- I.V. 4 ys 6 ms  
MPS I; severe  
cervical spine  
stenosis and  
occipitocervical  
instability
- Tetraplegia
- Hydrocephalus  
treated by  
ventricular  
derivation.



- I.V. posterior occiput-cervical decompression and occipital cervical stabilization by two rods, screws at occipital level and hooks at cervical level.

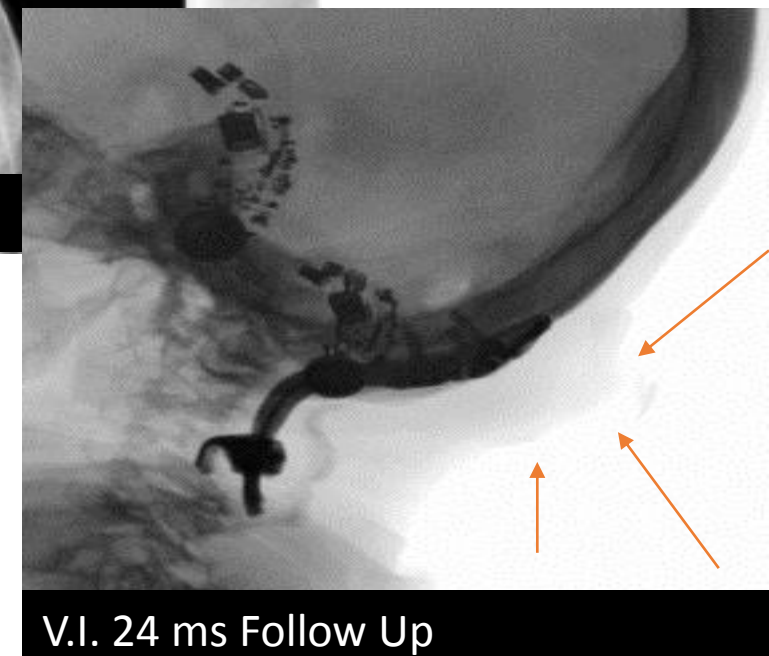
- **As there was no dedicated cervical pediatric instrumentation we used the smallest adult rigid instrumentation available**



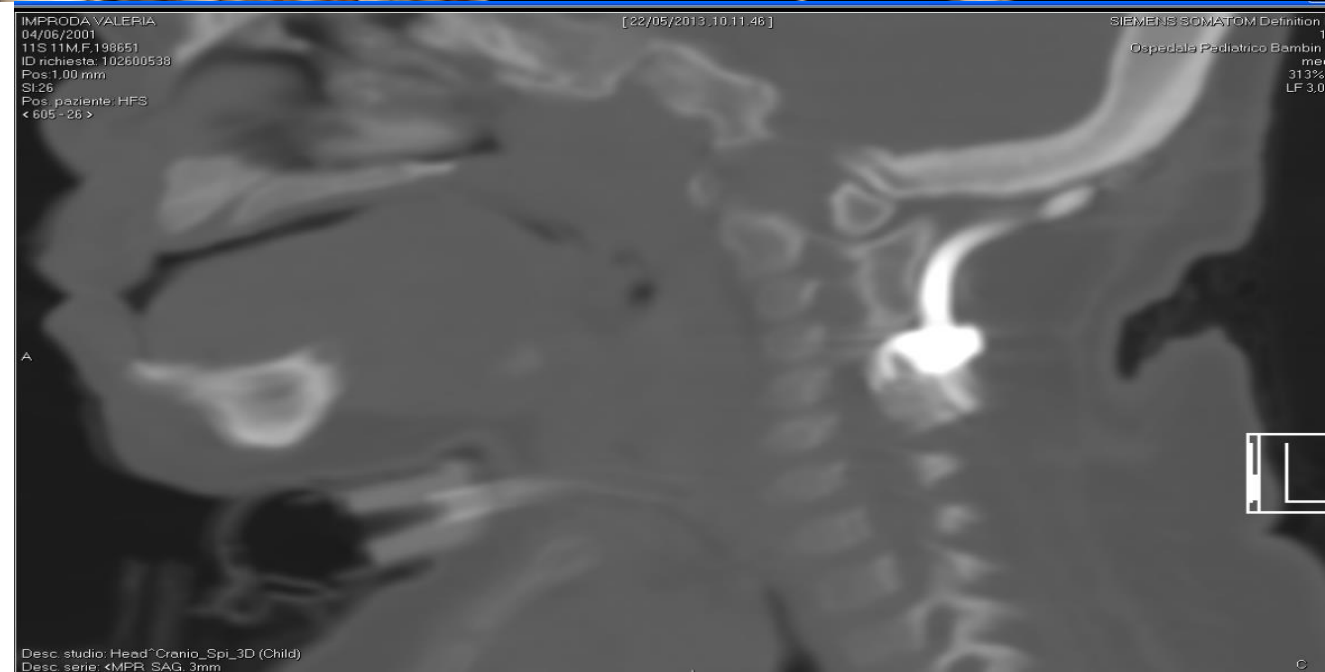
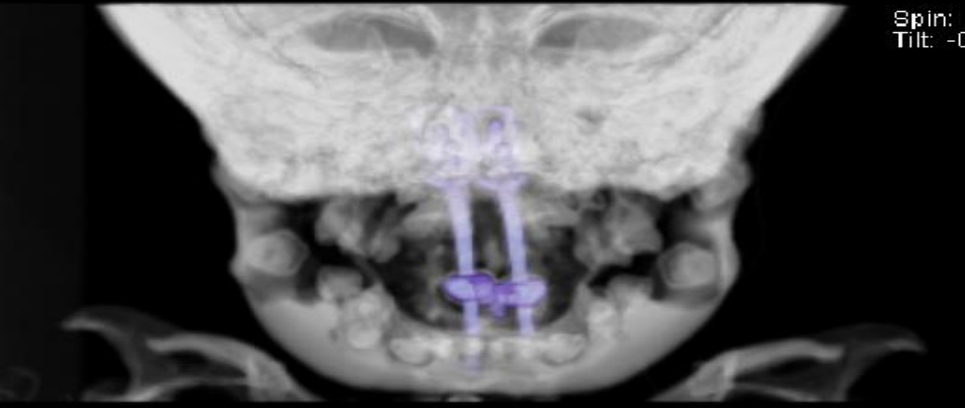
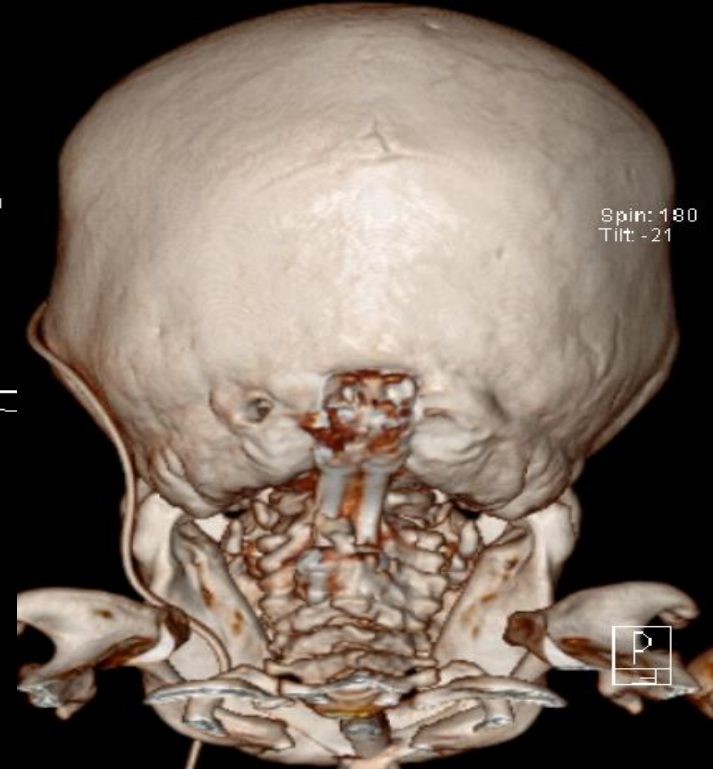
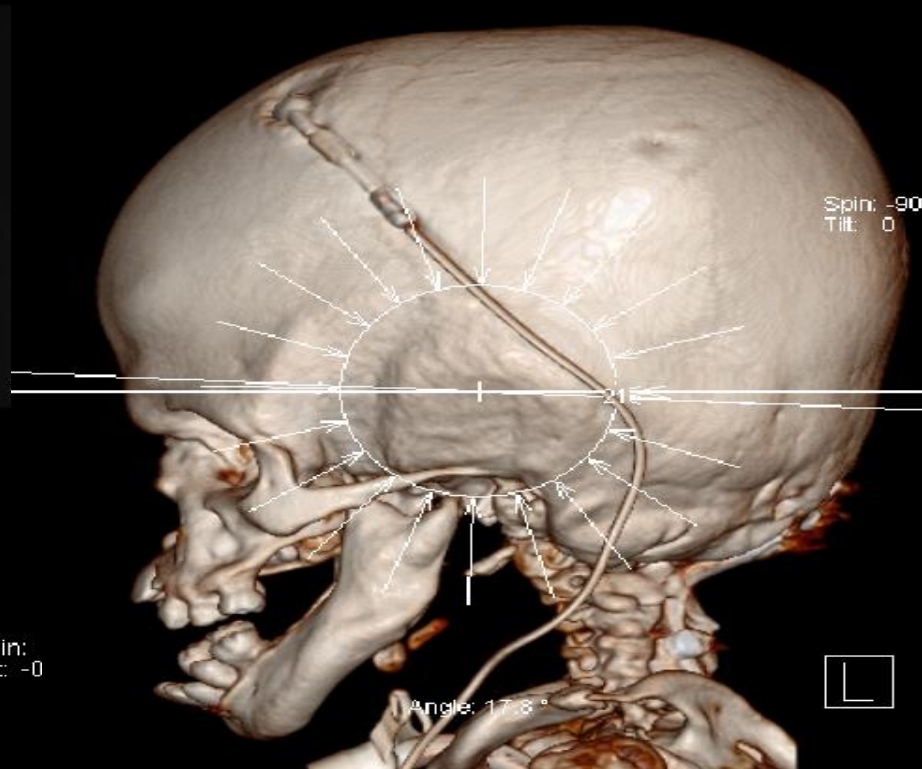
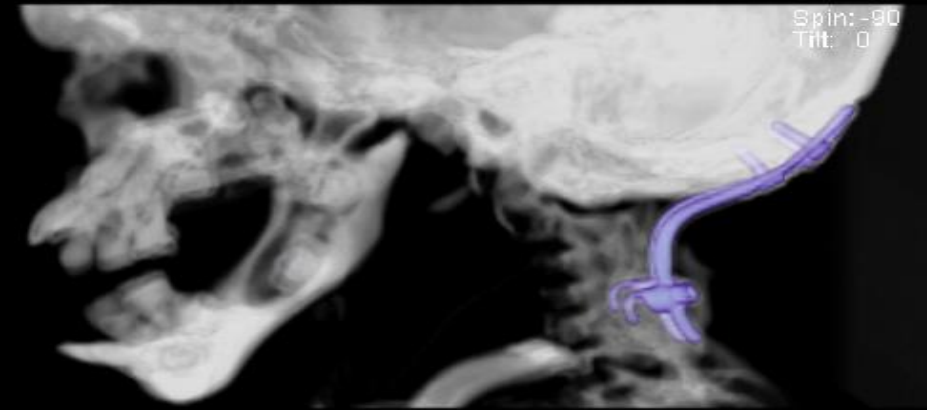


Recovery of the neurological status after surgery and at 6 month follow up she regained partial use of upper extremities.

At 24 months follow up she devoped soft tissue swelling in the occipital area due to reactive bursitis from protruding instrumentation. Bursitis was solved by anti inflammatory drugs.







At 11 years follow up patient occiput cervical arthrodesis is stable, she has no pain from protruding instrumentation and her neurologic status is stable

# Conclusion

- **Bracing is a viable treatment strategy in thoracic lumbar kyphosis and can obtain good clinical results at medium terms follow up even if kyphosis deformity remains in radiographs.**
- **Surgical treatment is effective in severe evolving cases both at cervical and thoracic lumbar level, main difficulties arose from unavailability of dedicated instrumentation in very young patient, as even smallest devices available are often too big**



# Thank You--Merci



***Rome,  
Saint Peter and  
Sant'Angelo  
Castle seen from  
Tiber river at  
sunset***

