

Complications in Neurofibromatosis

Michael G. Vitale, MD MPH

*Ana Lucia Professor of Orthopedic Surgery
Columbia University College of Physicians and Surgeons*

*Co-Director Division of Pediatric Orthopaedic Surgery
Chief of Pediatric Spine and Scoliosis Service
Medical Director, MSCH Initiative to Make Care Better
New York-Presbyterian Morgan Stanley Children's Hospital*

-Disclosures-

Michael G. Vitale, MD MPH

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POSNA – BOD

IPOS- Chairman

Some Relevant

Neurofibromatosis 1

- One of the most **common** genetic disorders
- Affects **1/3,000** people
- **Autosomal dominant** loss of function mutation → deficiency of the **neurofibromin** tumor suppressor protein
- High incidence of **scoliosis: 10%-60%**

Diagnostic Criteria For NF

Box 1. Diagnostic criteria for neurofibromatosis type 1

More than six café au lait spots measuring at least 15 mm in adults and 5 mm in children

Two or more neurofibromas of any type or one plexiform neurofibroma

Freckling in the axillary or inguinal regions

Optic glioma

Two or more Lisch nodules (ie, iris hamartomas)

A distinctive bony lesion, such as sphenoid wing dysplasia, or thinning of the cortex of a long bone with or without pseudarthrosis

A first-degree relative with NF-1, as suggested by these criteria

Neurofibromatosis 1

- **Broken into two distinct subgroups**
 1. **Non–Dystrophic**
 2. **Dysplastic (Dystrophic)**

Non-Dystrophic Neurofibromatosis

- Also undergo **rapid progression**
- Considered more **similar to idiopathic scoliosis curves**
- May **evolve into dysplastic type**

Conversion of non dystrophic to dystrophic curves

Athanasios I. Tsirikos
Asif Saifuddin
M Hilali Noordeen

**Spinal deformity in neurofibromatosis
type-1: diagnosis and treatment**

- **Risk of conversion of non-dystrophic curves into dysplastic curves**
- **Reported as high as 80% in patients diagnosed before 7 yo**
- **25% in patients diagnosed after age 7**

Dysplastic Neurofibromatosis

- Associated with **skeletal dysplasia**
- **Early and relentless progression**, even after **bone growth has ceased**
- **Curve progression** even *after* **arthrodesis**
- **Short, sharp curves**
- **Poor bone stock, osteoporosis**

Radiographic Features of Dysplastic Curves- Durrani

■ Modulation of Spinal Deformities in Patients With Neurofibromatosis Type 1

Abubakar A. Durrani, MD,* Alvin H. Crawford, MD,* Sambhu N. Chouhdry, MD,*
Asif Saifuddin, FRCR,† and T. R. Morley, FRCSt

- Described **nine specific radiographic features** associated with dystrophic scoliosis
 1. Rib penciling
 2. Vertebral rotation
 3. Posterior vertebral scalloping
 4. Anterior vertebral wedging
 5. Lateral vertebral scalloping
 6. Vertebral wedging in either the sagittal or coronal plane
 7. Spindling of the transverse process
 8. Widened interpedicular distance
 9. Enlarged intervertebral foramina
- Curves with **three** or more features **progressed 12° annually** vs **5°** in curves with fewer than three features

Decreased BMD in NF

RESEARCH REVIEW

AMERICAN JOURNAL OF
medical genetics PART
A

Skeletal Abnormalities in Neurofibromatosis Type 1: Approaches to Therapeutic Options

Florent Elefteriou,^{1*} Mateusz Kolanczyk,^{2,3} Aaron Schindeler,^{4,5} David H. Viskochil,^{6,7} Janet M. Hock,⁸ Elizabeth K. Schorry,⁹ Alvin H. Crawford,¹⁰ Jan M. Friedman,¹¹ David Little,^{4,5} Juha Peltonen,^{12,13} John C. Carey,^{6,7} David Feldman,¹⁴ Xijie Yu,⁸ Linlea Armstrong,¹¹ Patricia Birch,¹¹ David L. Kendler,¹⁵ Stefan Mundlos,^{2,3} Feng-Chun Yang,^{16,17} Gina A\giostratidou,¹⁸ Kim Hunter-Schaedle,¹⁸ and David A. Stevenson^{6,7}

Scoliosis Associated with Neurofibromatosis

Alvin H. Crawford, MD, FACS^{a,*}, Jose Herrera-Soto, MD^b

^a*Cincinnati Children's Hospital, 3333 Burnet Avenue, ML 2017, Cincinnati, OH 45229-3039, USA*

^b*Orlando Regional Medical Center, 86 West Underwood Street, Orlando, FL 32806, USA*

Decreased Bone Mineral Density in Neurofibromatosis-1 Patients with Spinal Deformities

T. Illés¹, V. Halmai¹, T. de Jonge^{1,2}, and J. Dubousset²

¹Department of Orthopaedics, Medical and Health Sciences Center, University of Pécs, Hungary; and ²Service de Chirurgie Infantile, Hôpital Saint-Vincent-de-Paul, Université René Descartes, Paris, France

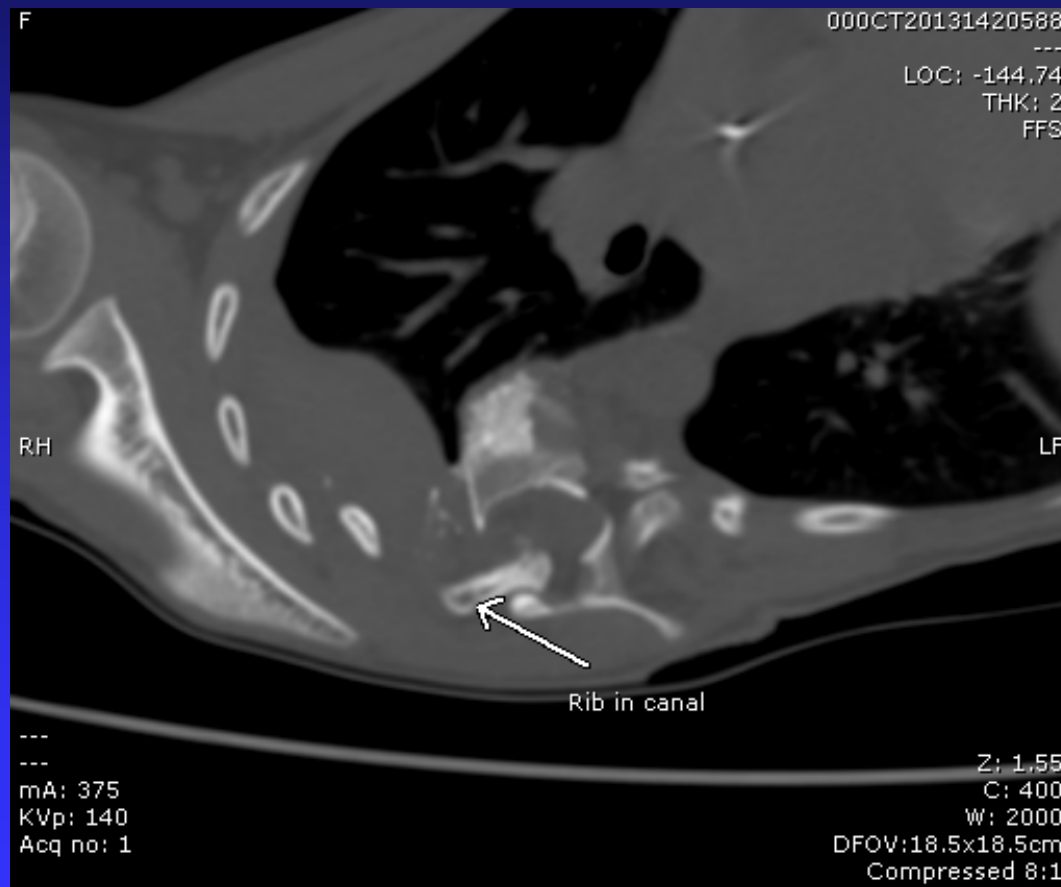
Neurofibromatosis Spine

Whole-Spine Magnetic Resonance Imaging in Patients with Neurofibromatosis Type 1 and Spinal Deformity

Manoj Ramachandran, MRCS, Athanasios I. Tsirikos, MD,* Joshua Lee, MRCS,* and Asif Saifuddin, MRCP, FRCR†*

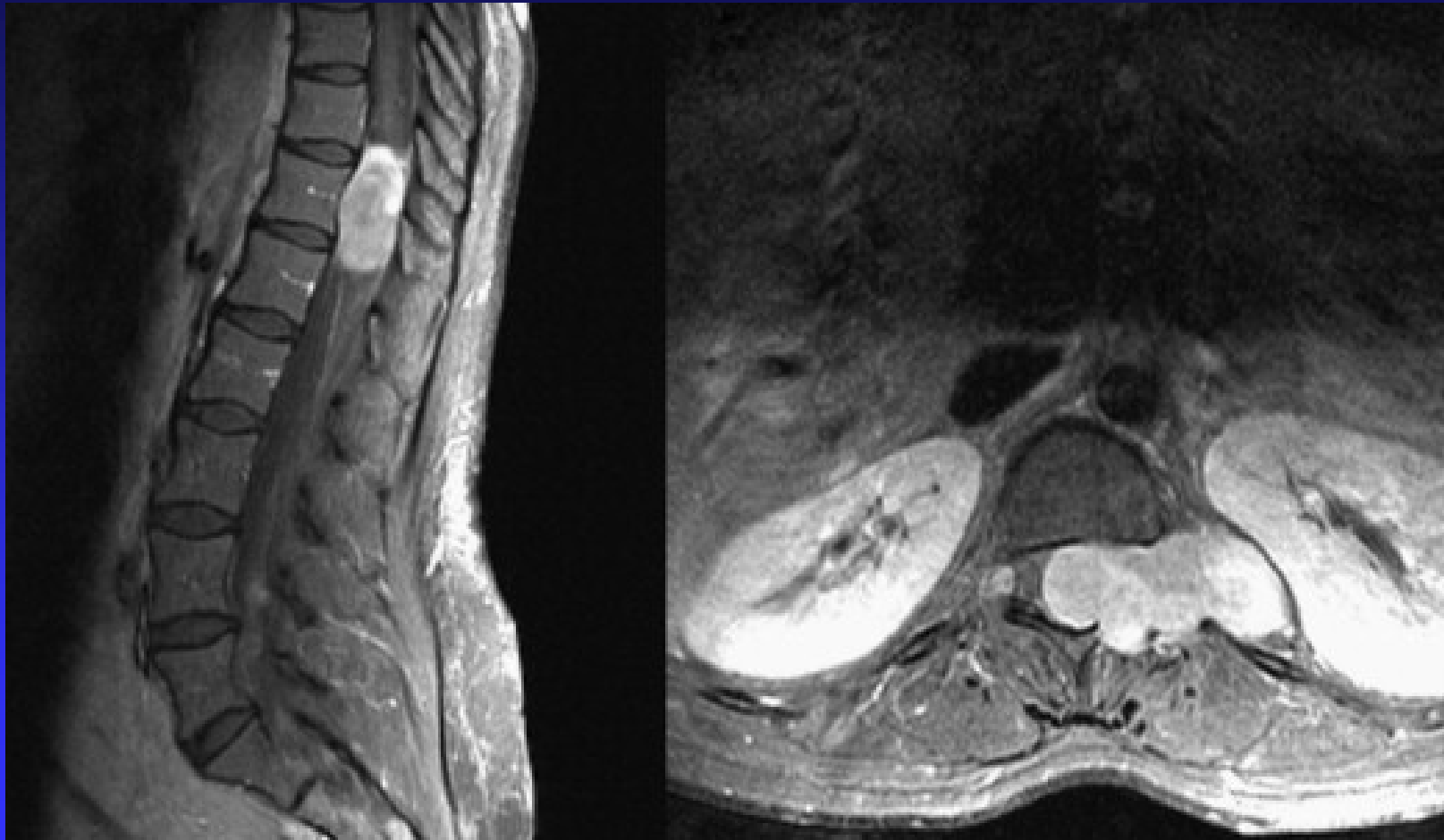
- Associated with **neurofibromas** adjacent to the vertebrae
 - **43.7%** in dystrophic; 25% in nondystrophic
- Essential to examine relationship of ribs and spinal canal!

Rib in Canal



Courtesy David Skaggs, MD

Dumbbell Neurofibroma



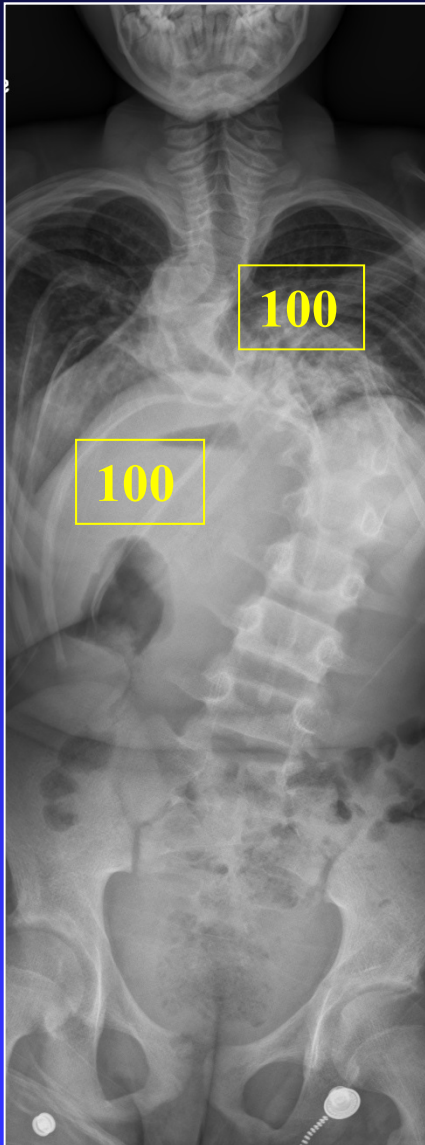
Complications Rates in NF Scoliosis

Study	Dural Tear %	Pseudarth. %	>10° Progr. Post Fusion %	Infx. %	Other (Specified)
Li et al. 2009	15.8	5.2	10.5	-	5.2 (distal hook dislodgement)
Wilde et al. 1994	-	-	52.0	-	12.0 (neurologic post-op injuries)
Tiziana et al. 2012	-	23.5	-	-	5.9 (proximal screw loosening)
Levy et al. 2015	5.9	5.7	-	10.2	5.7 (neuro compromise) 4.2 (hardware failure)
AIS Rates for Comparison	0.2 (Levy et al. 2015)	5.0 (Weiss & Goodall 2008)	-	1.4	0.8 (neuro compromise) 1.1 (hardware failure)

Case Presentation

KN

Patient KN: Neurofibromatosis 1



12yo M
NF Type 1
Neurologically intact

Scoliosis in NF:

**Modulates quickly and
unpredictably**

Associated with:

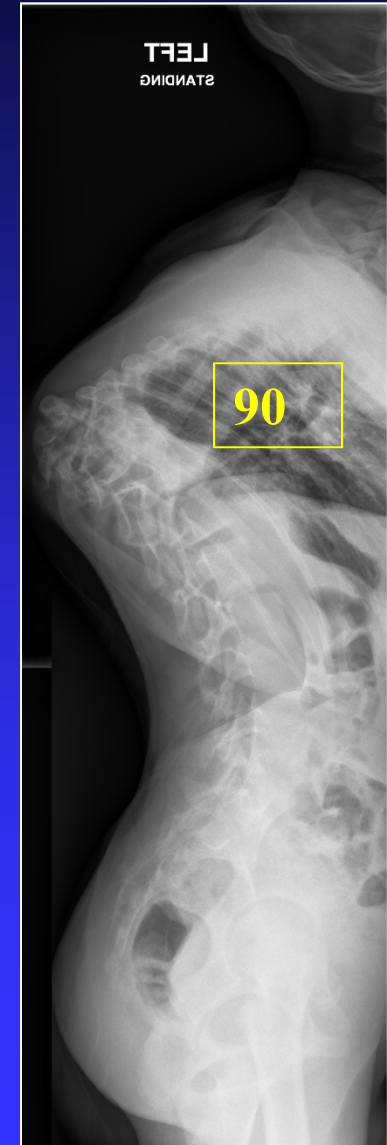
Sharp Angulation in all planes

Severe Rotation

Dural Ectasia

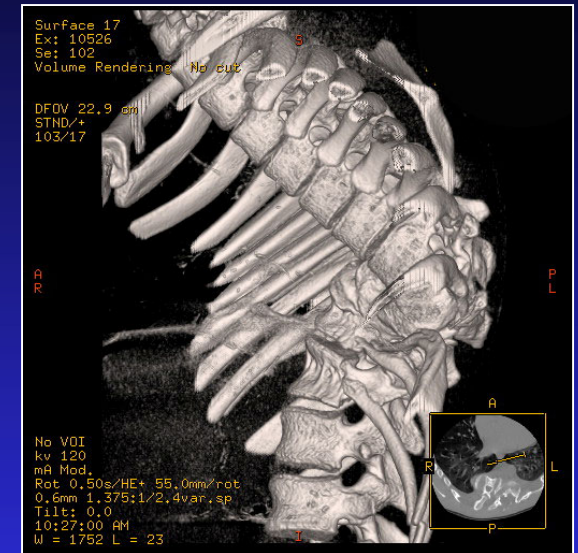
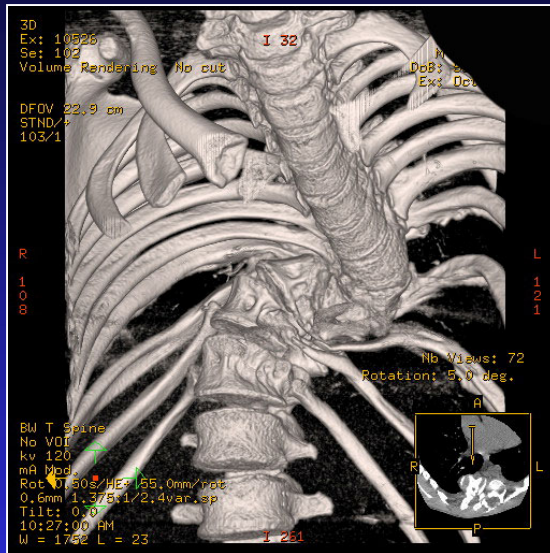
Pencilng of ribs

Foraminal Neurofibromas



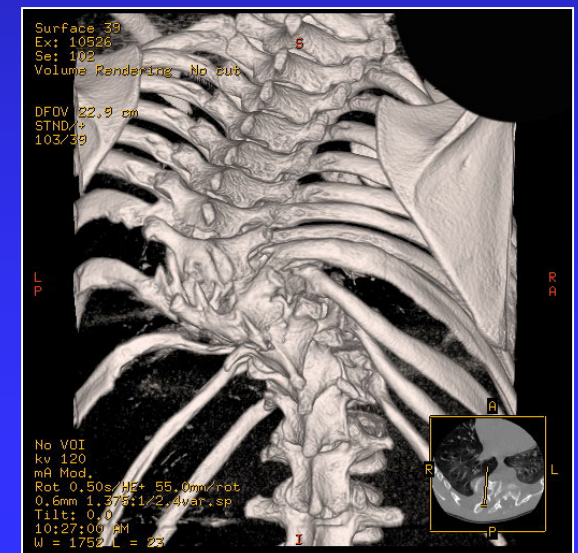
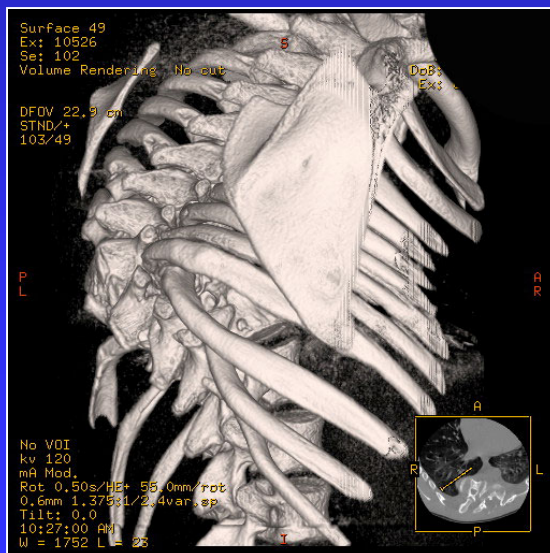
Patient KN: CT 10/26/2009

Severe
Kyphoscoliosis
Sharp Angulation at
T9



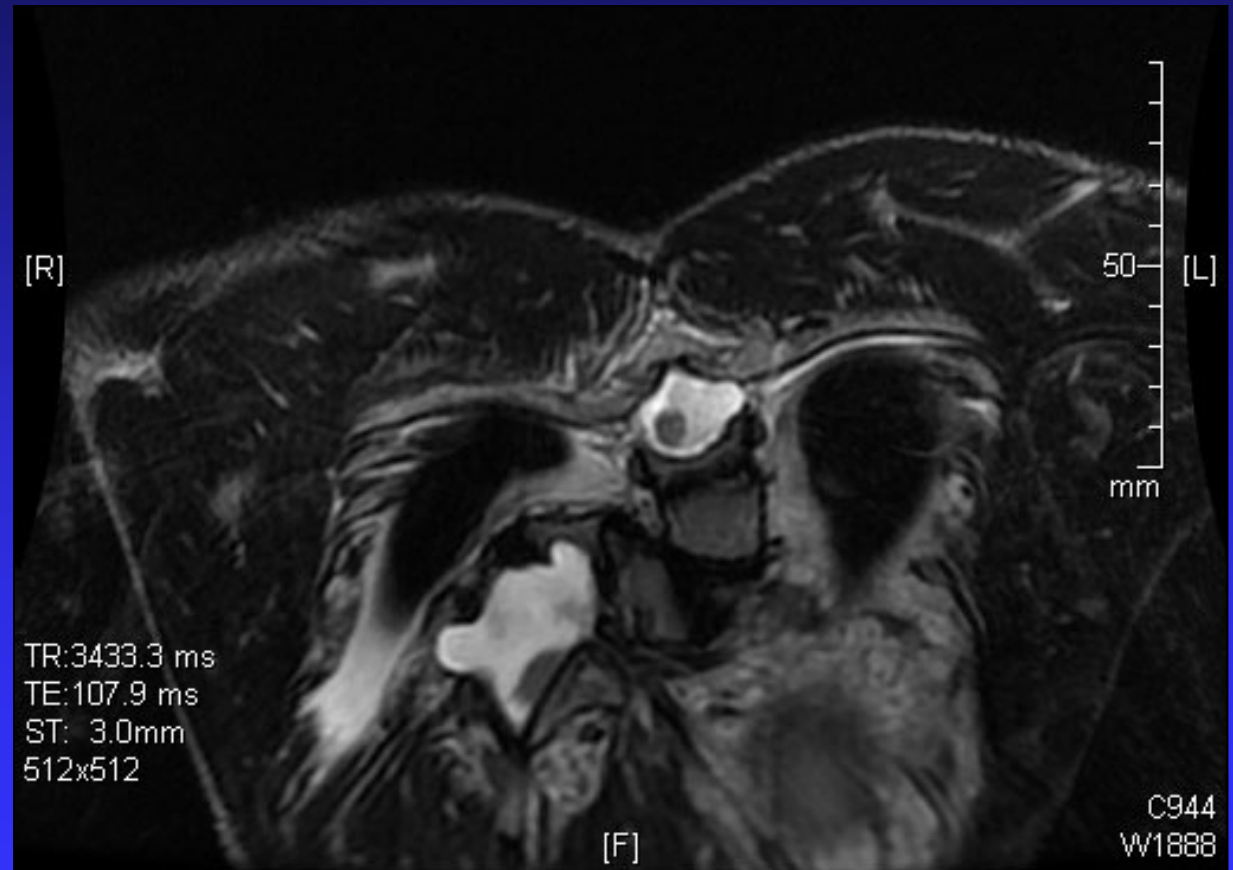
“Segmentation
Anomalies” of T8-
T11

Wispy pedicles at
apex



Patient KN: MRI

- Wide central canal at T8-T11
- Extensive dural ectasia



Surgical Options

1. Fusion in situ
2. Anterior Posterior Instrumentation Fusion
3. Posterior Only Approach
4. Use of Halo in Combination with Above
5. Choice of Rods?

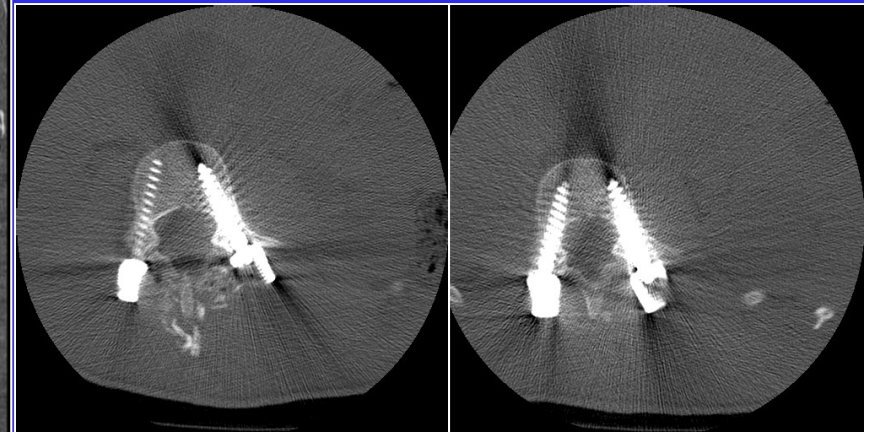
Patient KN Plan: Posterior-Posterior Staged Procedure

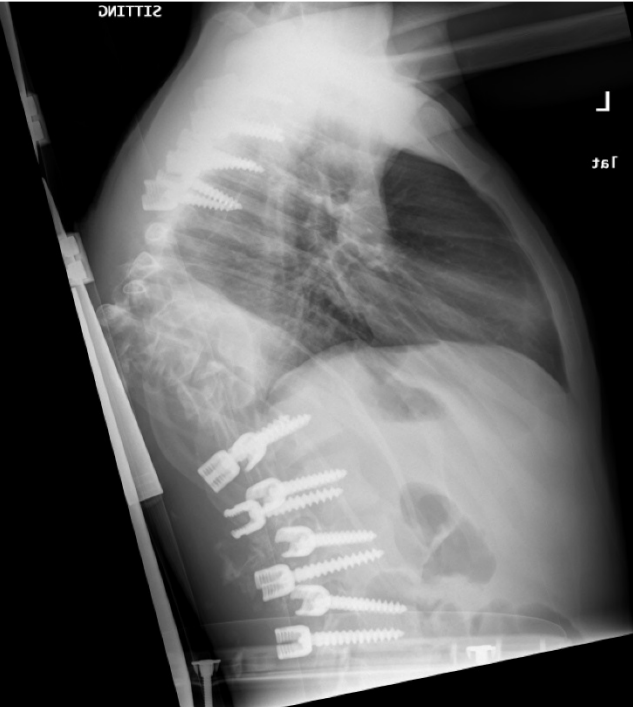
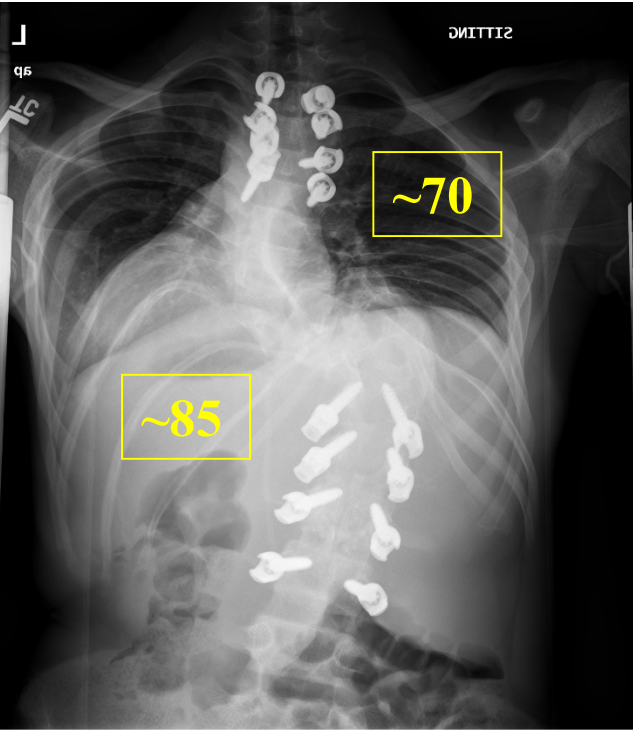
Stage 1 of 2: 10/25/2010

- Placement of Pedicle Screws: T2-T5, L1-L5
- Multiple SPO
- Application of Halo
- SSEPs/MEPs w/o Change
- Complications: Dural leaks with Screw Placement



CT: 11/4/2010





Patient KN

s/p 3 Weeks Halo Traction

Pre-op for Stage 2



Patient KN Plan: Posterior-Posterior Staged Procedure

Stage 2 of 2: 11/10/2010

- **Vertebral Column Resection – T9**
- **PSIF – T2-L4**
- **BMP + Extensive Autograft**
- **SSEPs/MEPs w/o Change**
- **Complications: CSF Leak Closed Primarily**

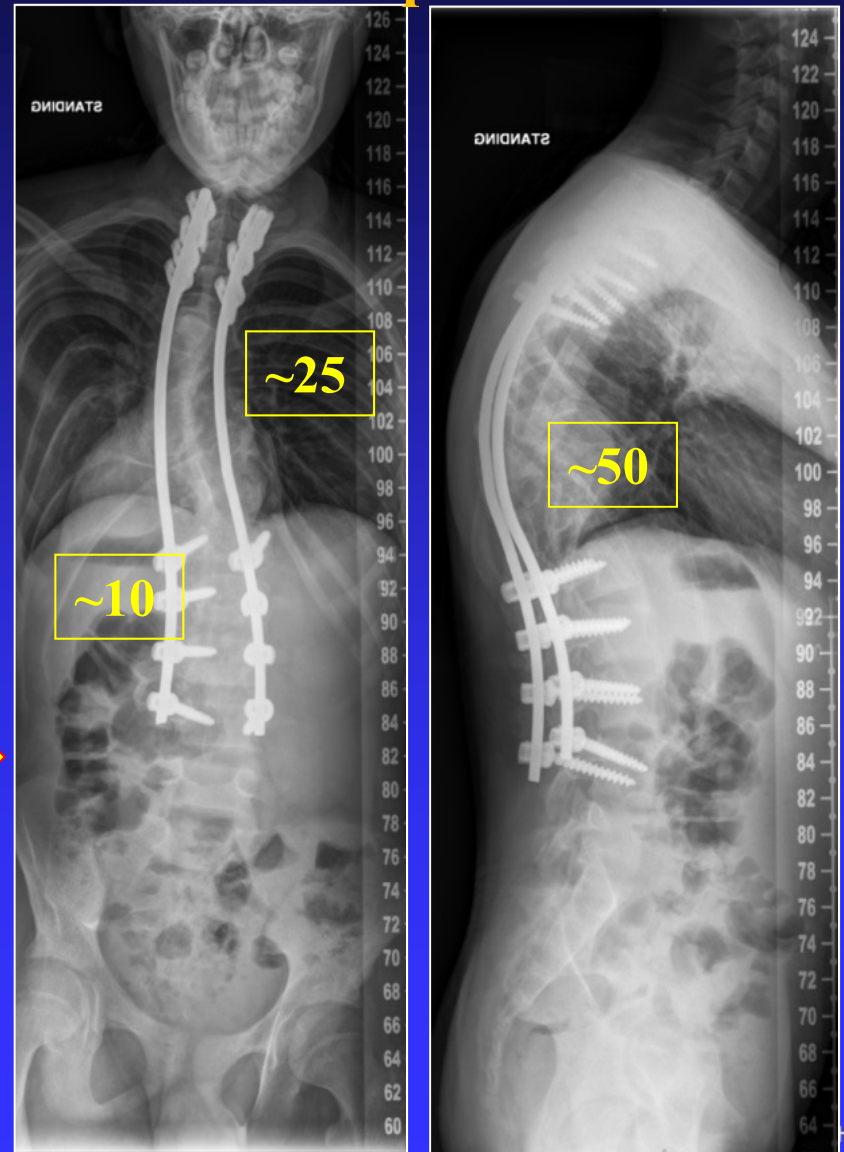
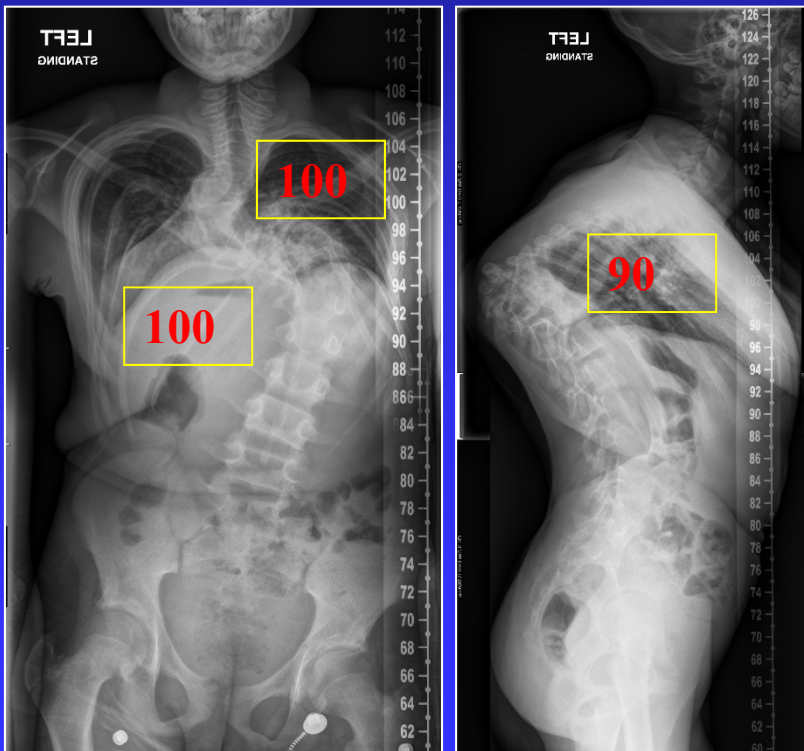


Post-op Stage 2

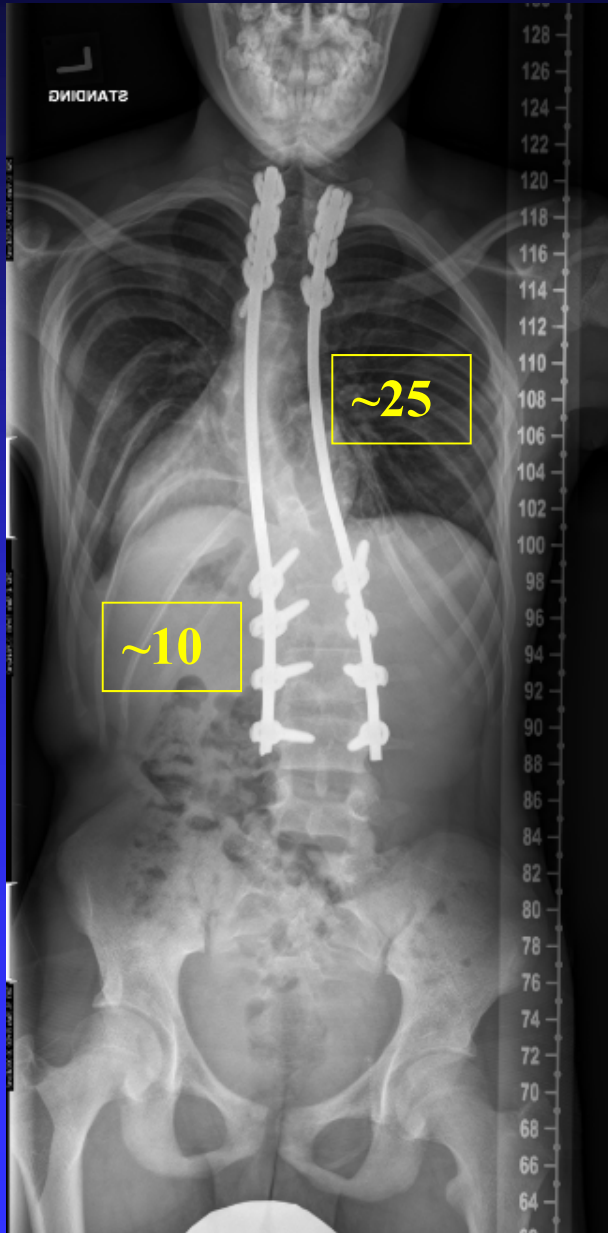
Patient KN: 6 Months Later

Post-Op 12/2010

- 4/2011: Doing very well
- No longer in PT → playing baseball and basketball



Patient KN: 72 Months Later

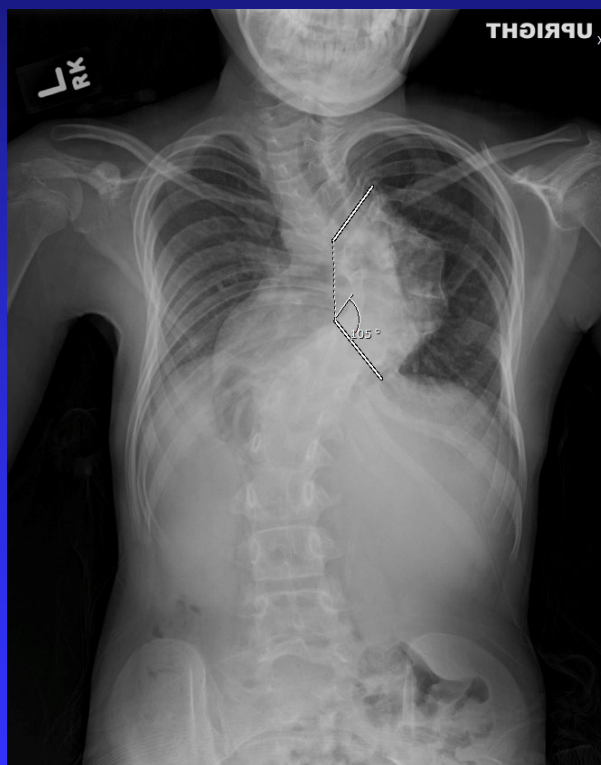


No change in curve

Is Anterior
Support
Necessary?



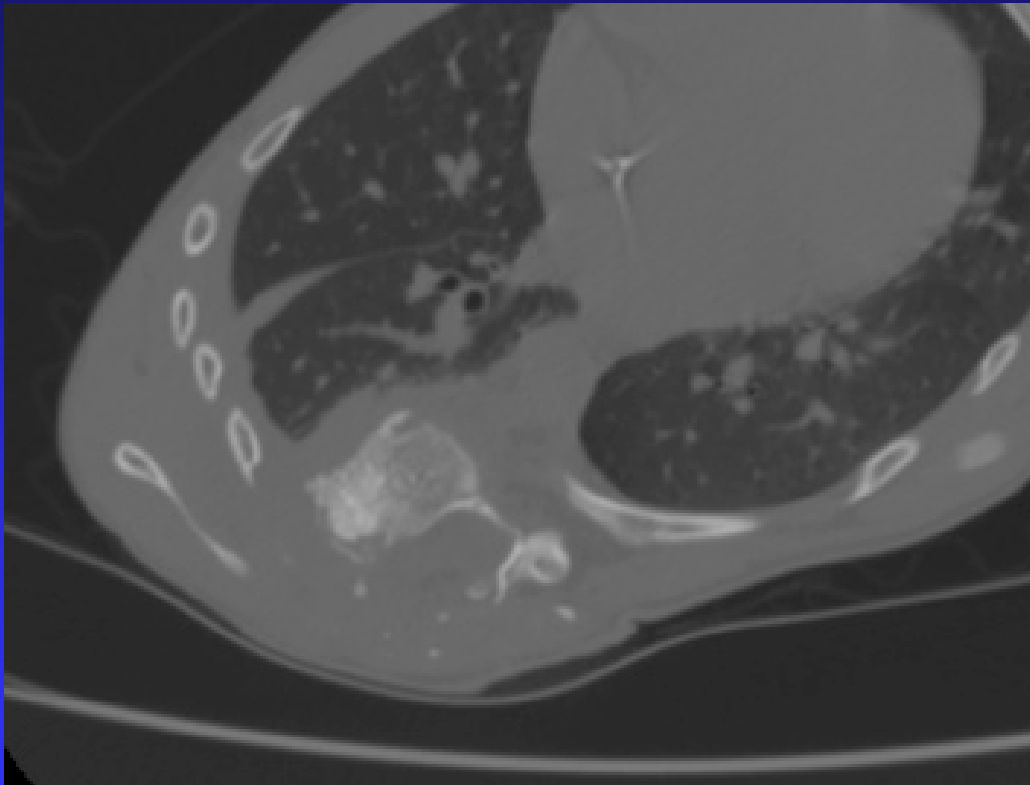
12 yo girl, neurofibromatosis 124 deg scoliosis



26

Courtesy David Skaggs, MD

Apical 4 levels no lamina or pedicles



12 yo girl, neurofibromatosis

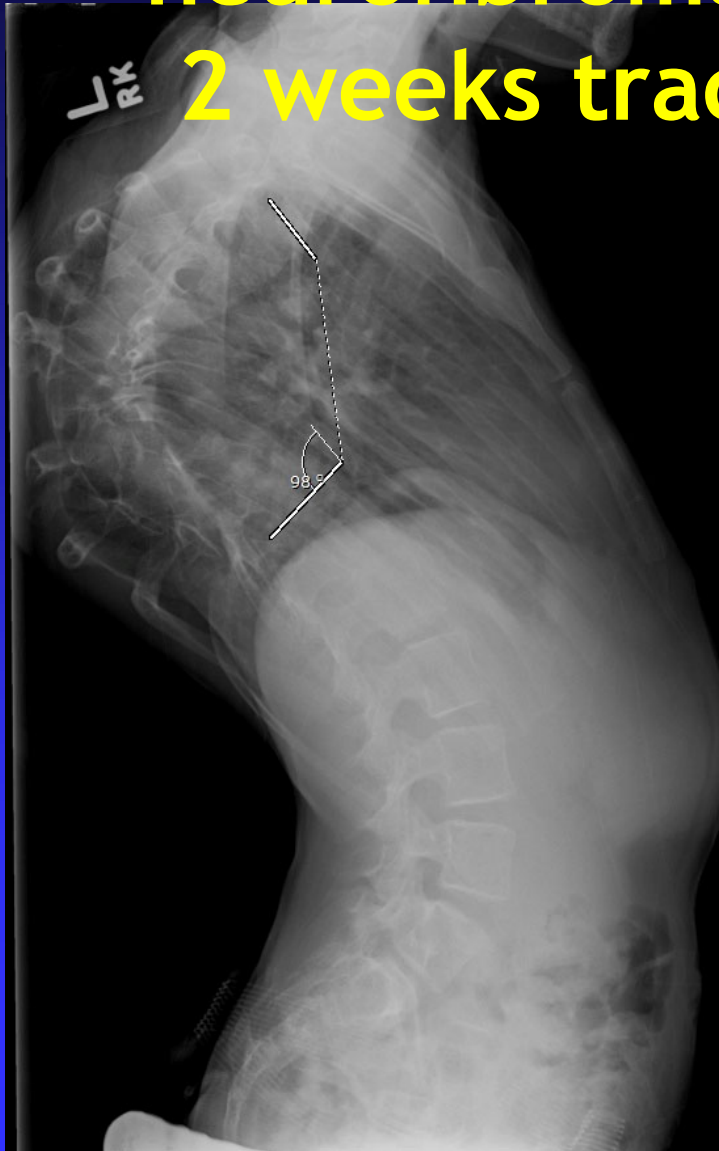
Anterior Release, 2 weeks tra 74 deg n



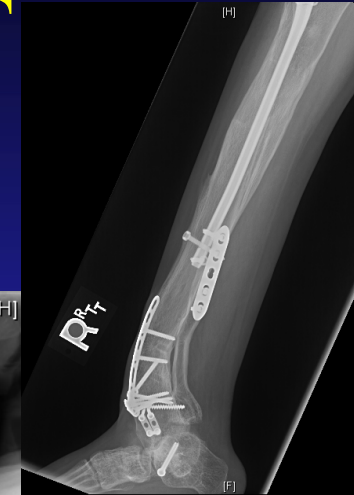
12 yo girl, neurofibromatosis 2 weeks traction



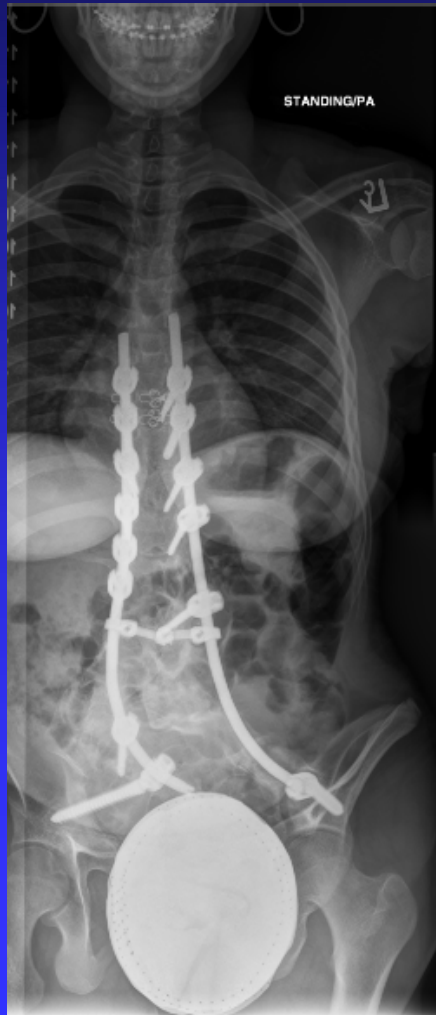
**12 yo girl,
neurofibromatosis
2 weeks traction**



AK: 12 year old with NF



12 year old with NF 6 years post op; BMP



NF Scoliosis

- **Difficult cases with high risk**
- **? Necessity of anterior fusion in modern era of implants**
- **High complications**



Thank You

Michael G. Vitale, MD MPH

mgv1@columbia.edu

