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Syndromic Cervical Spine

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Lisbon, Portugal
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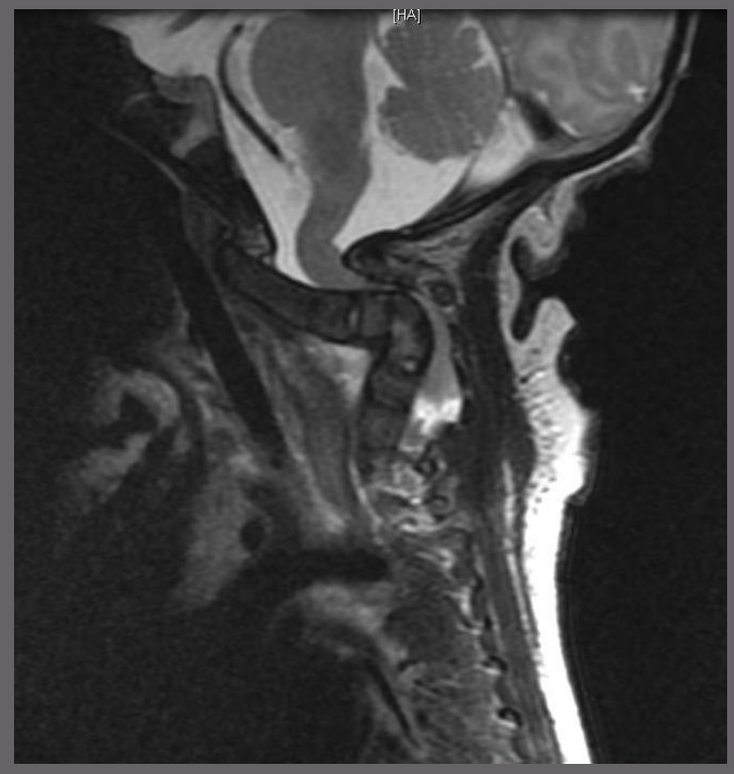


Disclosures

- I could not bring myself to present a book report summarizing > 200 syndromic skeletal dysplasias affecting the cervical spine

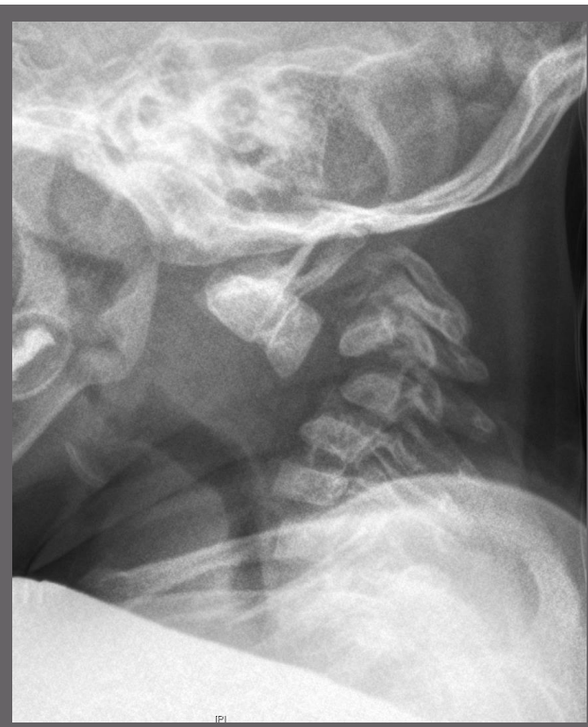
TABLE 235-2 Congenital Abnormalities Affecting the Subaxial Cervical Spine

Disorder(s)	Spinal Pathologic Findings	Additional Findings	Radiographic Appearance
Klippel-Feil syndrome and Klippel-Feil variant	Cervical fusion at any level(s),[17] adjacent instability[18]; occipitalization of the atlas[19]	Classic triad: short neck, low hairline, and limited neck mobility due to congenital cervical spine fusion[17]; Chiari I malformations[19]	Abnormal bony architecture, with normal bone quality[19]
VATER/VACTERL syndrome	Hypoplastic vertebrae, or hemivertebra	Vertebral anomalies, anal atresia, cardiac abnormalities, tracheoesophageal fistula, esophageal atresia, renal and radial dysplasia[20]	Hypoplastic vertebrae may be evident on chest radiograph
Down's syndrome (trisomy 13)	Occipitoatlantal subluxation(40%-50%), atlantoaxial subluxation (10%-30%)[112-116]; rarely, subaxial instability[117,118]	Intellectual disability; characteristic facies; single palmar crease	Vertebral subluxation
Neurofibromatosis type 1 (NF1)	Progressive cervical kyphosis—extensive surgical reconstruction may be necessary[119]	Neurofibromas; café-au-lait spots; axillary or inguinal freckles; optic nerve glioma; Lisch nodules; long-bone abnormalities	Scalloping of the vertebral column; paraspinal neurofibromas; expansion of the foramina, remodeling of the facet joints[119]
Achondroplasia	Spinal stenosis greater at foramen magnum than at subaxial cervical spine or thoracolumbar spine[120-122]	Most common form of dwarfism; characterized by disproportionate shortening of the proximal ends of limbs relative to the trunk[120-122]	Spinal stenosis
Goldenhar's syndrome (oculoauriculovertebral dysplasia)	Vertebral hypoplasia; failure of segmentation (usually, cervical spine at C3-C4); failure of vertebral formation (thoracolumbar spine)[123,124]	Clinically heterogeneous constellation of symptoms characterized by spinal defects, hemifacial microsomia, and epidermoid appendages as well as craniofacial, cardiac, renal, gastrointestinal, and ophthalmic abnormalities	Vertebral hypoplasia
Kniest's syndrome	Osteopenia; atlantoaxial instability; odontoid hypoplasia; cervical hypoplasia and instability; kyphoscoliosis of the thoracolumbar spine[125-129]	Characterized by abnormal facies as well as large, stiff joints with contractures[125-129]	Osteopenic vertebral bodies
Morquio's syndrome	Atlantoaxial subluxation with cord compression; odontoid dysplasia[130-133]	Cardiopulmonary and neurological complications; often does not manifest until 2-6 yr of age[130-133]	Subluxation ±cord compression
Osteogenesis imperfecta	Pathologic fractures; type IV associated with craniovertebral junction abnormalities[134,135]	Type I mild, no long bone deformities; type II most severe, results in perinatal death due to in utero fractures; type III most severe form in patients who survive; type IV associated with moderate bone deformities and variable short stature[134,135]	Pathologic fractures
Larsen's syndrome	Hypoplastic vertebral bodies, dysraphism, hemivertebrae, wedged vertebrae, and midcervical subluxation; progressive kyphosis with progressive instability, myelopathy, and further segmental weakness[136-144]	Commonly associated with knee and hip dislocation, flattened facies, and cervical kyphosis[136-144]	Hypoplastic vertebral bodies, wedged vertebrae, subluxation, kyphosis
Spondyloepiphyseal and other skeletal dysplasias	Atlantoaxial instability, odontoid hypoplasia with cord compression, myelopathy[145,146]	Short-trunk disproportionate dwarfism[145]	Odontoid hypoplasia or os odontoideum

VATER/VACTERL, vertebral anomalies, anal atresia, cardiac defects, tracheoesophageal fistula and/or esophageal atresia, renal and radial anomalies, and limb defects.

Clinical pearl 1: Stall

- As long as possible
- Most cases of deformity/instability picked up incidentally or via screening
- Bracing allows moderate reduction, stability, and time for growth
- *Permits eventual stabilization with standard rigid internal fixation systems*



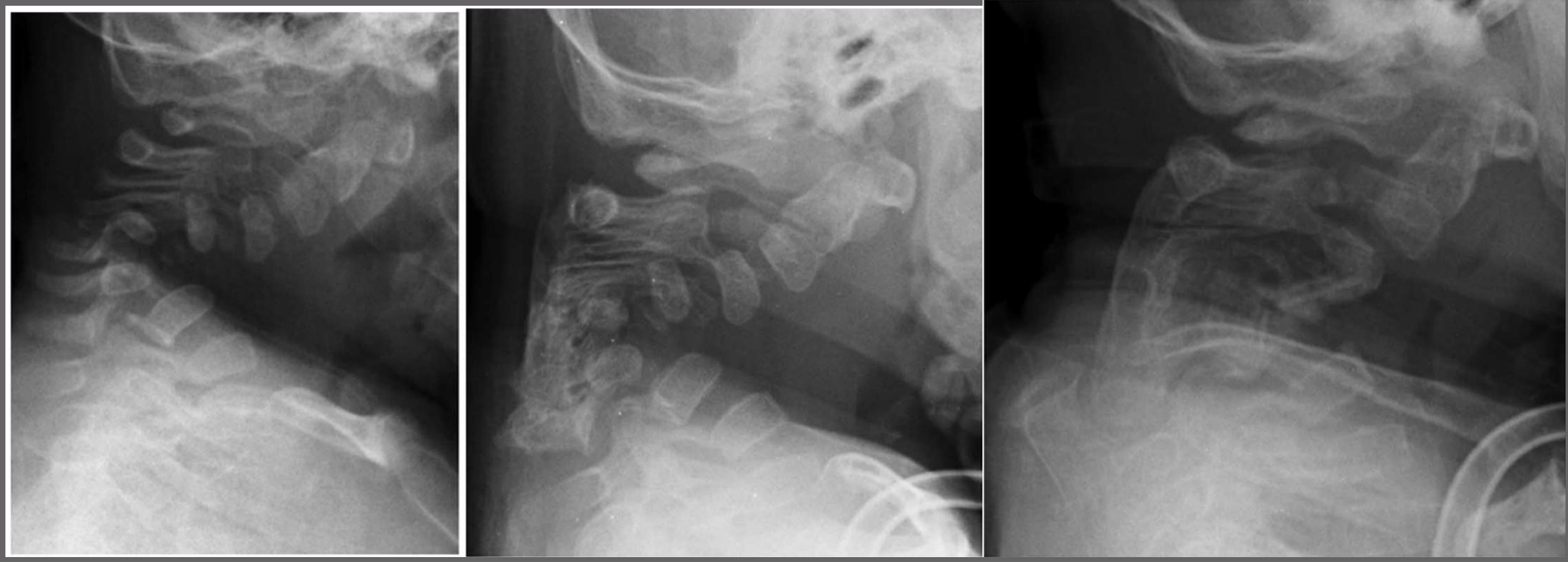
Clinical pearl 1: Stall

10	2.4	8.5	F		C4	118	Diastrophic dysplasia
11	0.5	4.3	M	×	C3	56	Conradi syndrome
12	0.8	6.4	M		C5	78	Larsen syndrome
13	1.0	8.8	M	×	C5	130	Larsen syndrome
14	18.3	51.1	F		C5	62	Type 1 neurofibromatosis
15	1.2	32.3	F		C4	87	Larsen syndrome
16	1.4	9.8	M		C4	30	Larsen syndrome
17	3.7	16.0	M		C5	40	Larsen syndrome
18	1.5	9.4	M		C5	41	Larsen syndrome
19	3.3	14.4	F		C5	108	Larsen syndrome
20	1.7	8.8	M	×	C4	37	Conradi syndrome
21	2.9	12.0	F	×	C6	18	Type 1 neurofibromatosis
22	12.4	44.6	F		C6	58	Gorham's disease
23	2.7	13.0	M		C4	90	Larsen syndrome
24	12.4	33.6	M	×	C4	49	Larsen syndrome

Martus et al. Spine 2011

Clinical pearl 1: Stall

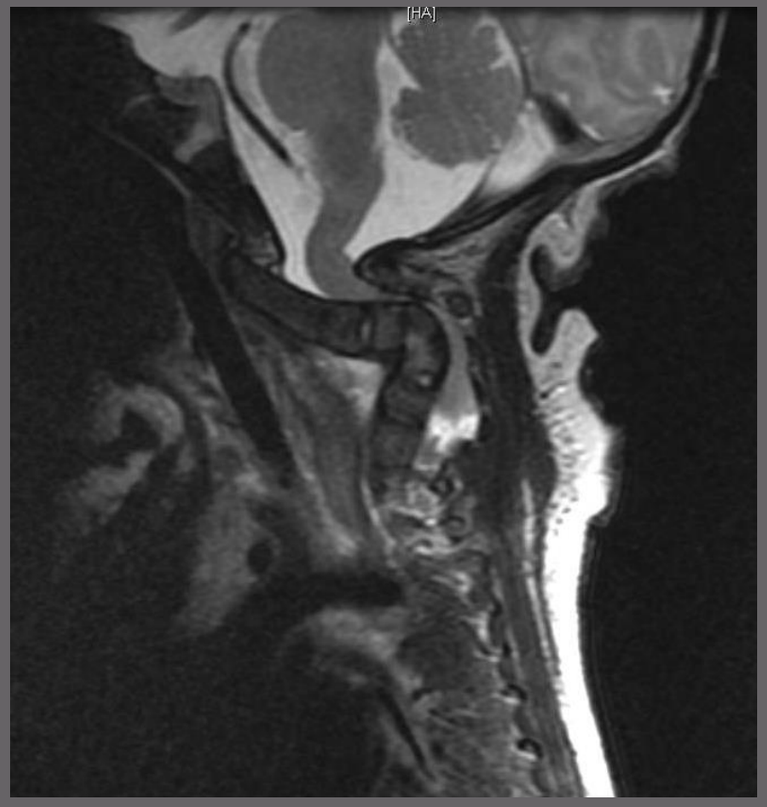
- 12/15 (80%) cases </= 3 y.o.; 6/15 (40%) </= 1.5 y.o.
- No rigid instrumentation used (wires/halo)
- Major complications in 9/15 (60%) of cases (quadriplegia, pseudo, etc)

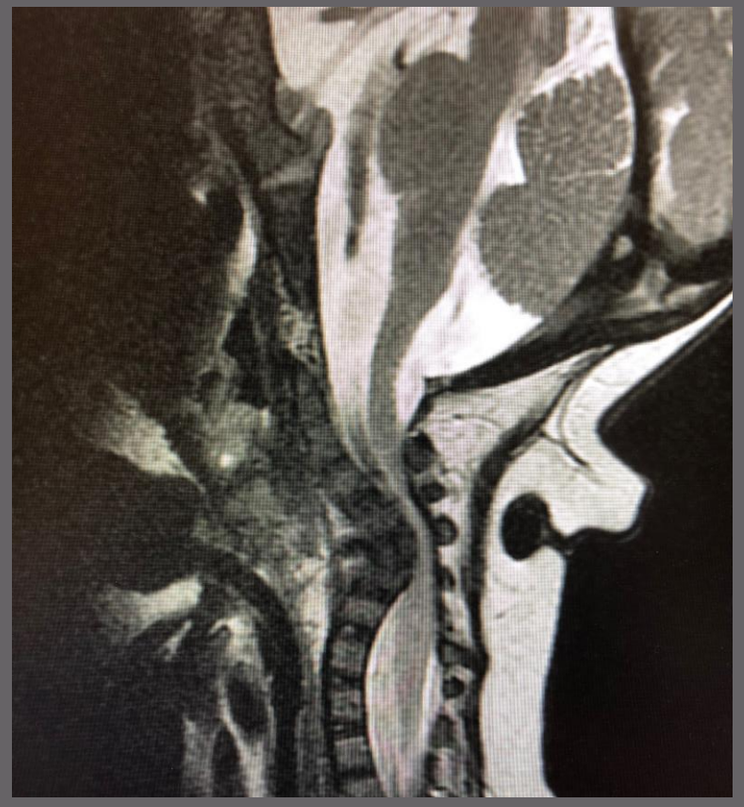


Martus et al. Spine 2011

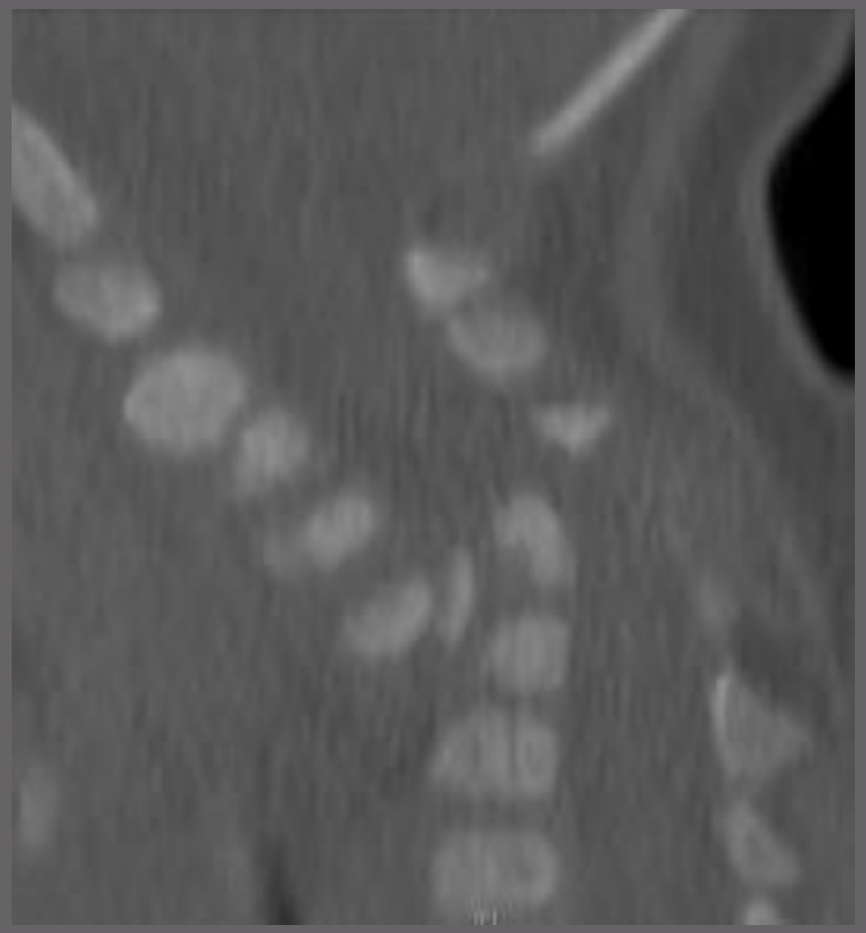
Clinical pearl 2: Use traction

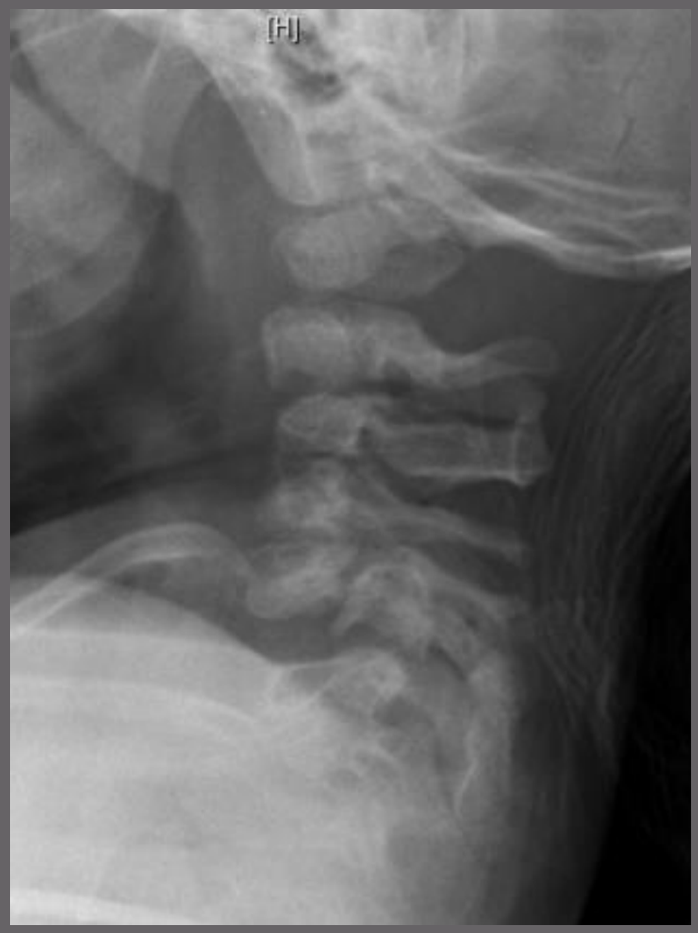
- Painful and barbaric for surgeons, patients, and families, *but it works*
- Can be done at any age
- Minimizes the chances of needing an anterior approach
- Safe: Permits assessment of neurological exam during reduction
- SSEPs/MEPs may not be monitorable in some cases

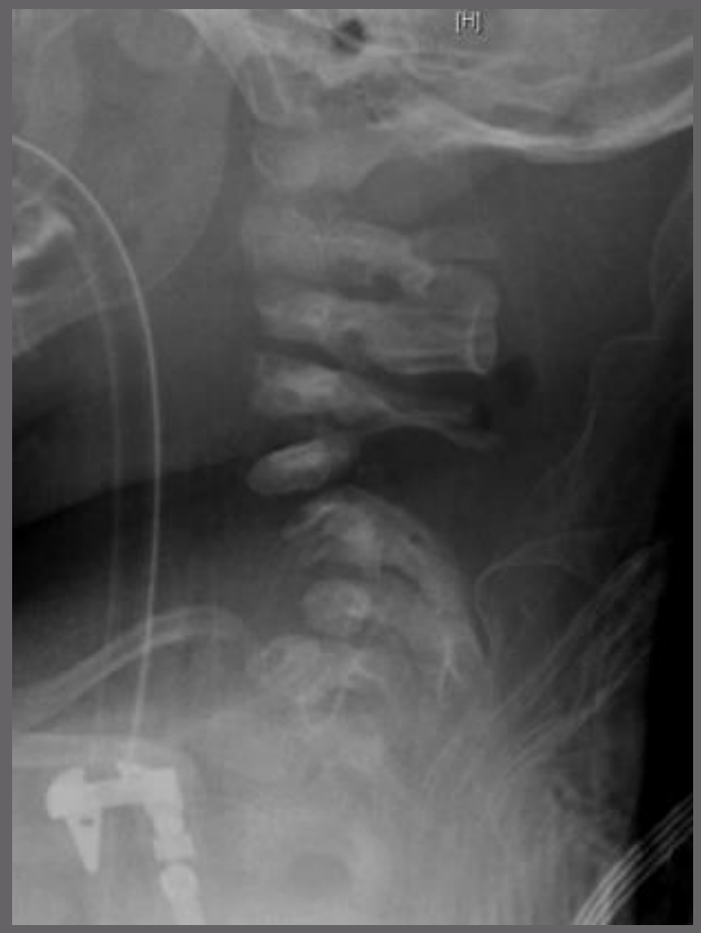




Clinical pearl 2: Use traction







- *No monitorable MEPs or SSEPs at baseline!*

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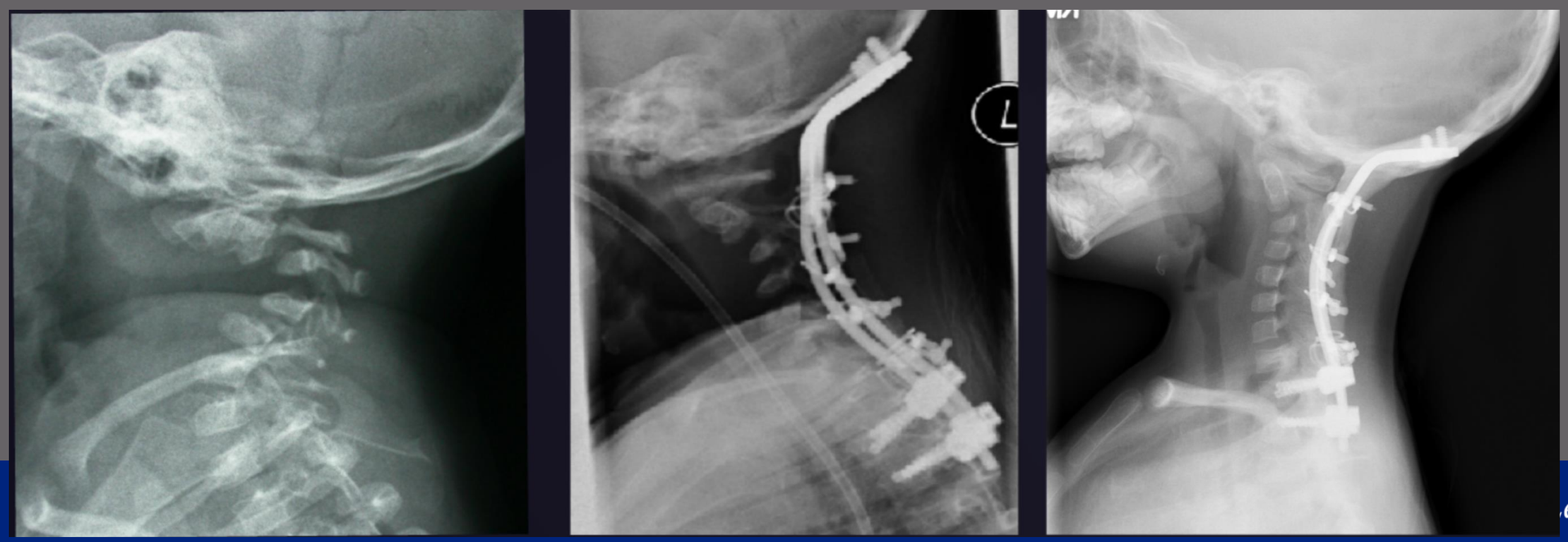
Clinical pearl 3: Start posterior

- More familiar approach with less risk
- Primarily involve kyphosis or swan neck deformities
- Most require occipital-thoracic instrumentation for adequate deformity correction
- Commercially available anterior instrumentation systems difficult in kids < 6-8 y.o.; Hand/OMF systems provide limited biomechanical strength
- In many cases (but not all), posterior alone will be sufficient



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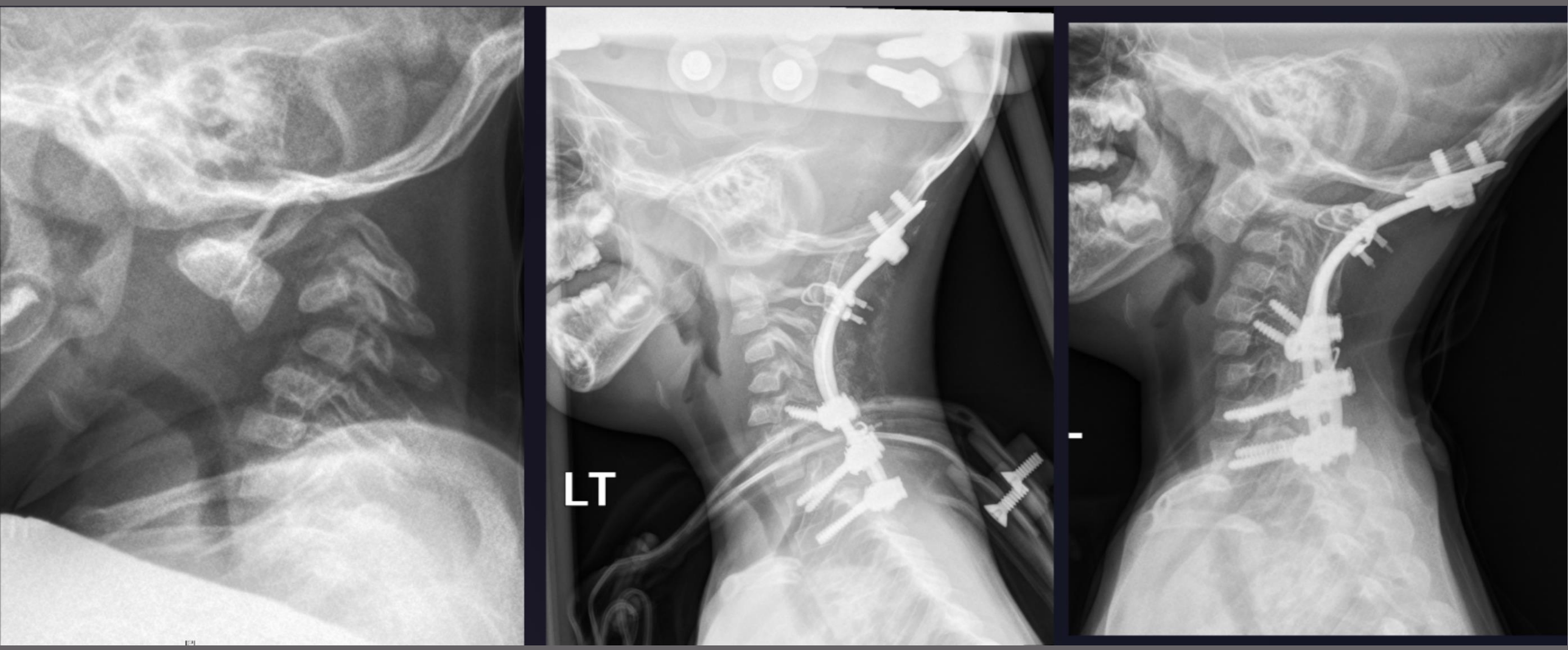
Clinical pearl 3: Start posterior



Fargen et al. JNS:Peds 2011

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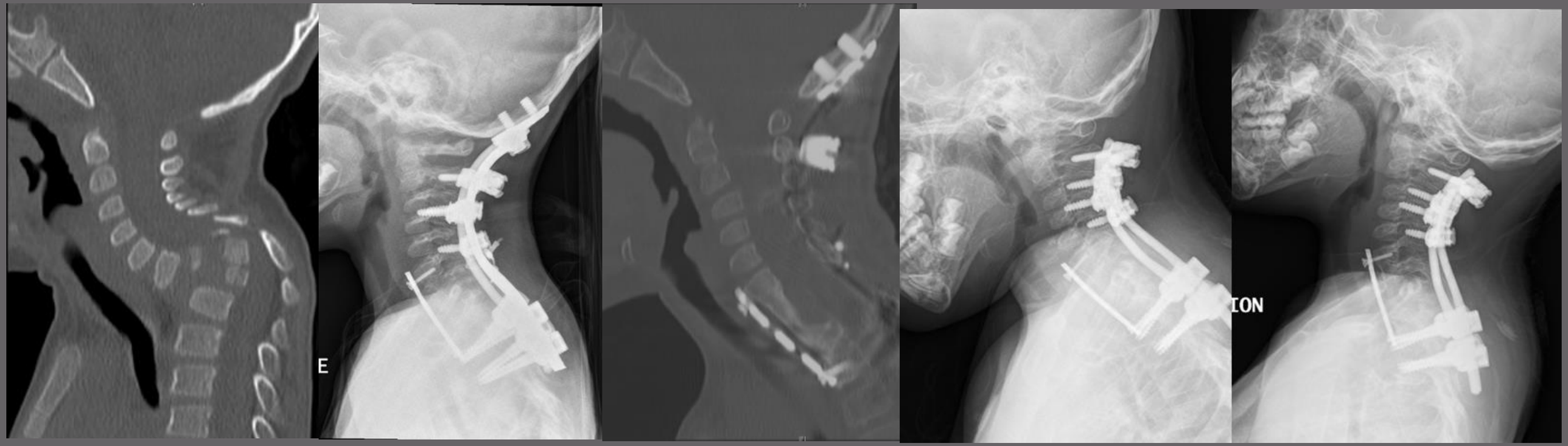
Clinical pearl 4: Instrument long/fuse short

- Fixation into occipital keel and thoracic pedicle screws often necessary
- Instability may not exist throughout entire cervical spine
- Must consider this at initial operation
- Disconnect above and below after solid fusion achieved where needed

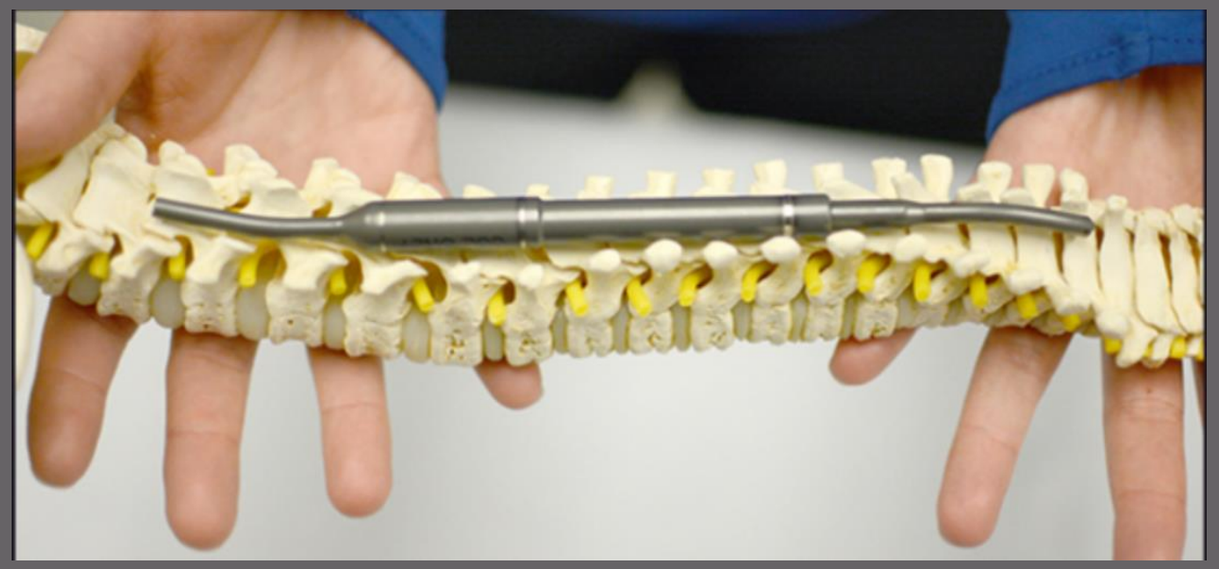


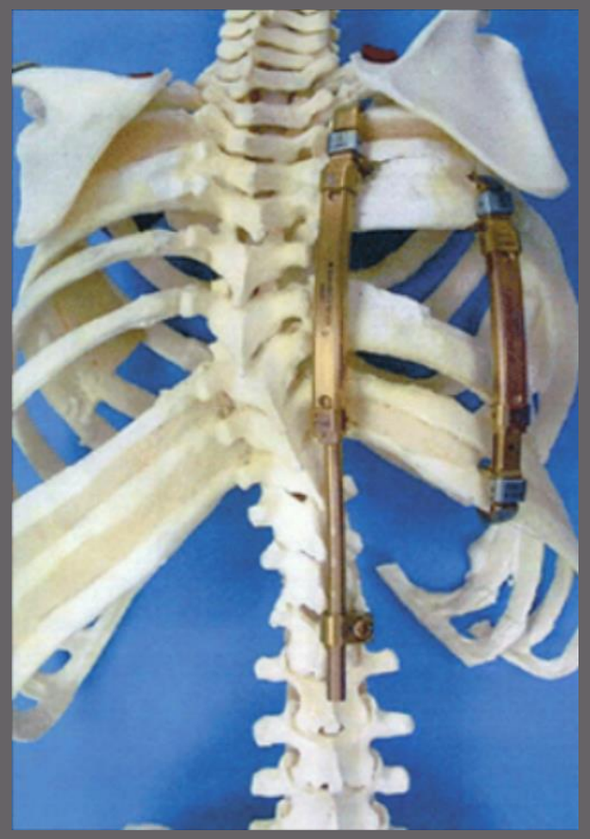
Clinical pearl 4: Instrument long/fuse short

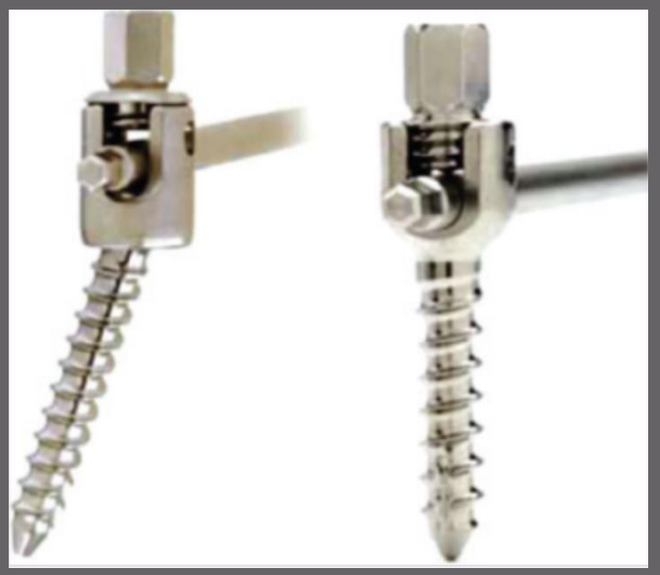
- Multicenter study investigating occipital-cervical-thoracic instrumentation and fusion in pediatric patients
- 13 syndromic patients with variety of etiologies
- Combination of anterior/posterior approaches
- 11/13 with preserved or improved neurological function in short term followup (mean 2.5 years)
- Longer term follow up: nearly 50% have required revision surgery

Future directions

- Need to do better with long constructs in young children
- Restricted growth and head movement
- VEPTR, MAGEC, other growing constructs too large for cervical spine
- Modified Shilla system?
- We need to think outside this galaxy. . .







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Future directions

And let these kids dance!

Thank you!

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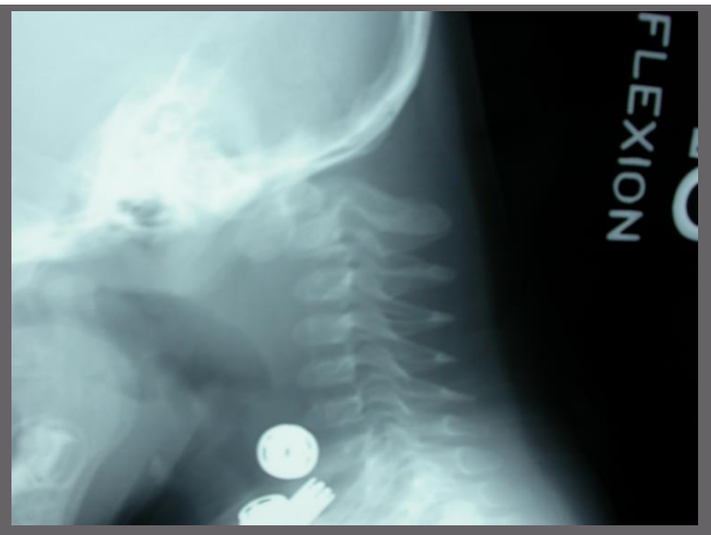
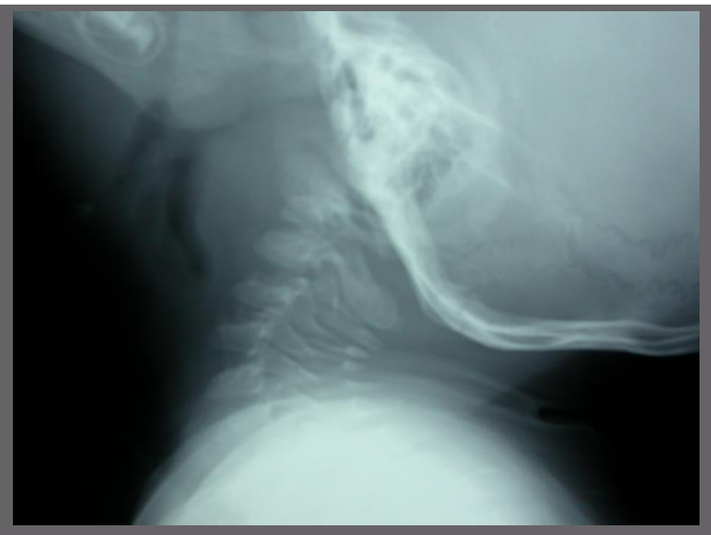
The syndromic cervical spine: common culprits

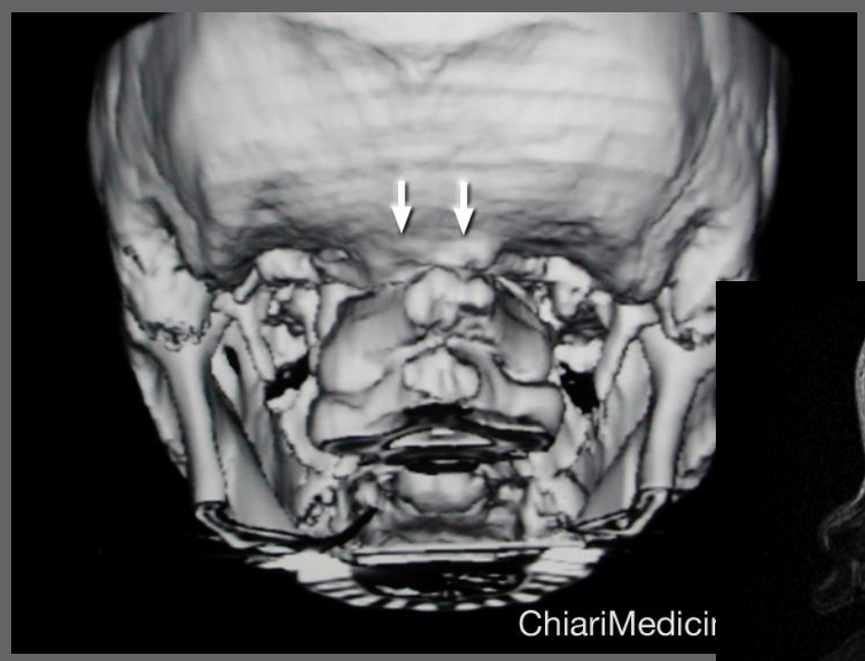
•Down syndrome	**Atlanto-occipital instability, atlantoaxial instability, rarely subaxial instability**
•Kniest's syndrome	**Atlantoaxial instability, odontoid and cervical hypoplasia and instability**
•Morquio's syndrome	**Atlantoaxial subluxation, odontoid dysplasia**
•Spondyloepiphyseal dysplasia	**Atlantoaxial subluxation, odontoid dysplasia**
•Klippel-Feil	**Cervical fusion at any level, adjacent instability, occipitalization of the atlas**
•VACTERL	**Hypoplastic or hemivertebra**
•Larsen's syndrome	**Hypoplastic or hemivertebra, wedged vertebra and midcervcal subluxation, progressive kyphosis**
•Achondroplasia	**Spinal stenosis greater at the foramen magnum than subaxial cervical spine**
•Osteogenesis imperfecta	**Pathologic fracture, type IV associated with craniovertebral junction instability**

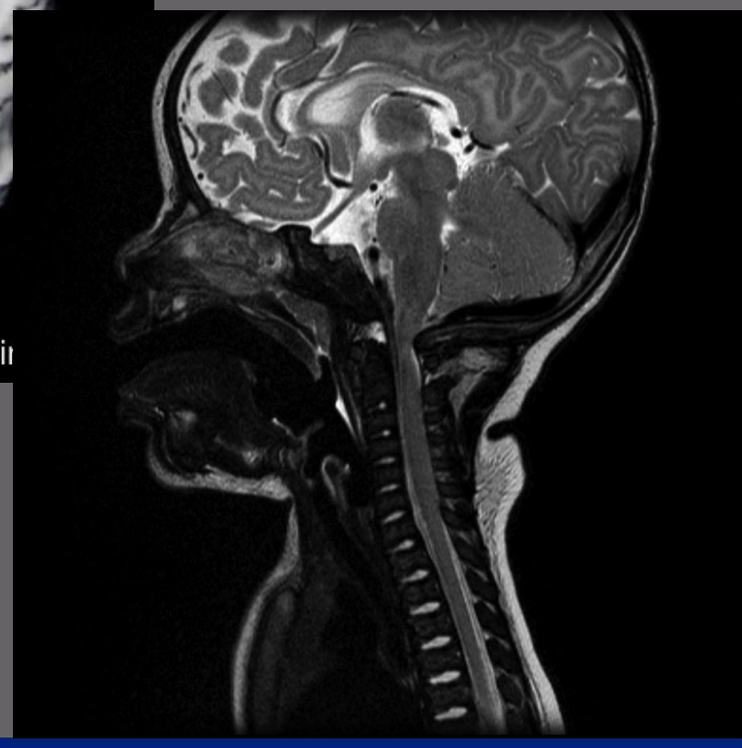
The syndromic cervical spine: Occiput-C1

- Atlanto-occipital instability
  - Seen in 40-50% of Down syndrome patients (flattened C1 articular “cup”)
  - Osteogenesis imperfecta
- Occipitalization of the atlas
  - Klippel-Feil
  - Goldnhar syndrome
- Foramen magnum stenosis
  - Achondroplasia
  - Common radiographic finding (60-70%), but less common clinical manifestations(10-35%)



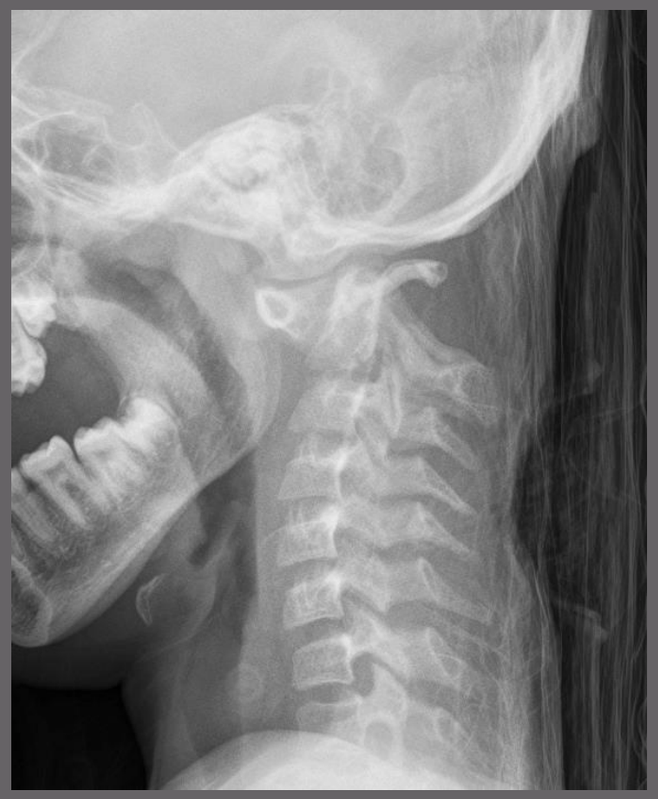


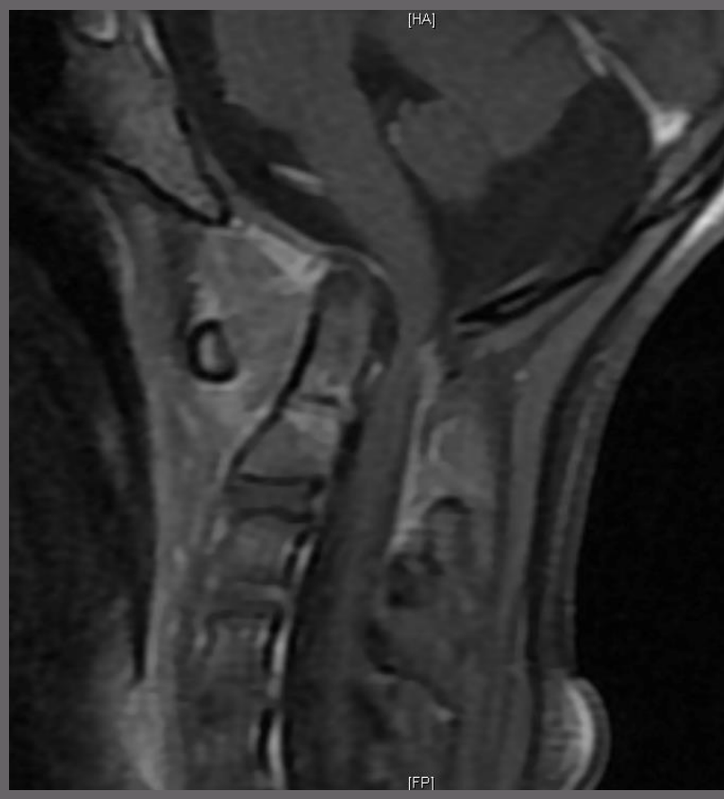


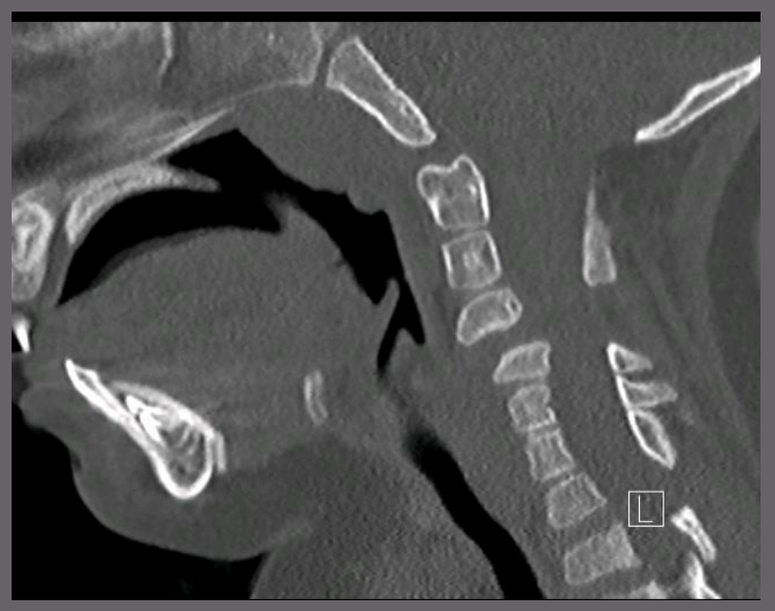


The syndromic cervical spine: C1-C2

- Atlantoaxial instability
  - 10-30% of patients with Down syndrome
  - Kniest's and Morquio's (42-90%) syndrome
  - Spondyloepiphyseal dysplasia
- Odontoid dysplasia and hypoplasia
  - Kniest's and Morquio's syndrome
  - Spondyloepiphyseal dysplasia

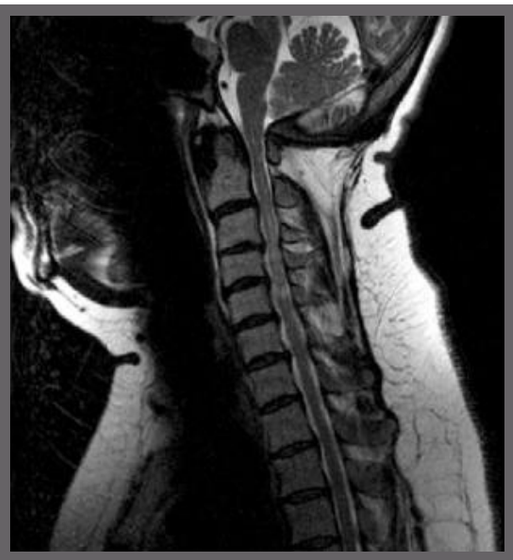


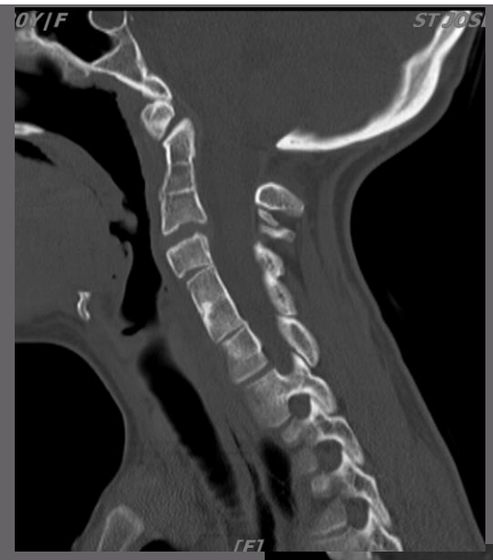


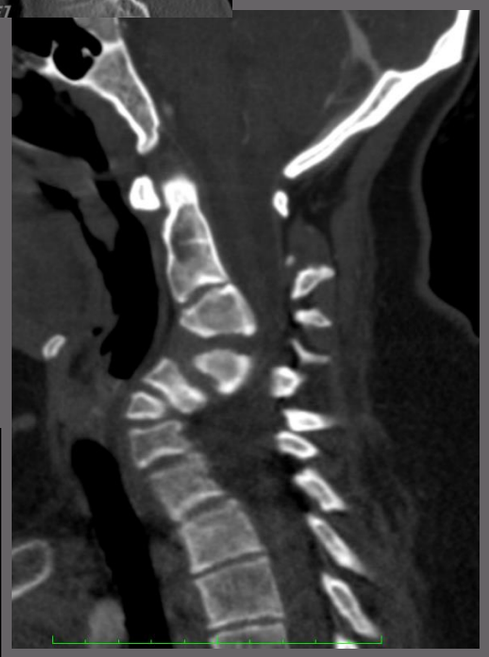


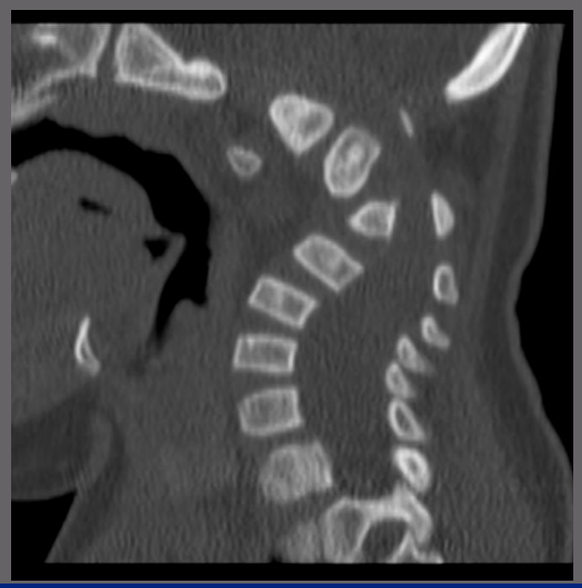
The syndromic cervical spine: subaxial cervical spine

- Subaxial stenosis
  - Achondroplasia
- Aberrant fusion and adjacent instability
  - Klippel-Feil
- Hypoplastic and hemiverteba
  - VACTERL
  - Larsen’s syndrome
- Pathologic fractures
  - Osteogenesis imperfecta









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The syndromic cervical spine: Diagnosis

- A-P, lateral and open mouth Xray
- CT scan with 3D reconstruction
- Cervical MRI
- Occiput-C1
  - CCI
- C1-2
  - ADI, PADI
- Subaxial
  - Flexion-Extension films to assess for reducibility when indicated