

# Early Onset Spinal Abnormalities, Treatment, and Complications in Loeys-Dietz Syndrome

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JOHNS HOPKINS  
M E D I C I N E

# Disclosures

Sara Fuhrhop: None

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Paul Sponseller:

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# Background

## Loeys-Dietz Syndrome (LDS)

- Autosomal dominant connective tissue disorder
- Mutations in TGF- $\beta$ R1 & TGF- $\beta$ R2 genes
- Characterized by vascular tortuosity and aggressive aneurysm formation, midline defects, and musculoskeletal abnormalities
- Early recognition



# Background

Spinal abnormalities have been reported and involve all aspects of the spine among patients of all ages:

- 19% cervical spine formation defects or instability
- 25% scoliosis
- 67% dural ectasia

# Objectives

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In LDS patients  $\leq 10$  years old:

- Quantify the prevalence of spinal abnormalities
- Report treatment and complications

# Methods

- Patient population
  - 42 patients diagnosed with LDS
  - Age  $\leq 10$  years (median  $6.8 \pm 3.5$ , range 1.3-10.5)
  - Average length of follow up 5.3 years
- Data collection
  - Cervical spine neutral, flexion, and extension x-rays
  - TL spine x-rays and CT
  - Some imaging modalities were not available for all patients, so results are reported as a fraction of those imaged.

# Results

## Cervical Spine

Cervical spine instability in 18 of 40 patients (45%)

- Atlanto-axial instability in 5 of 40 patients (12%)

Subluxation of C1-C2 > 3.5 mm in neutral position or on flexion/extension

- Subaxial instability in 14 of 40 patients (35%)

Subluxation of C2-C7 > 3.5 mm in neutral or flexion/extension

Mean age at diagnosis of instability 5.9 years, range 1 – 10 years



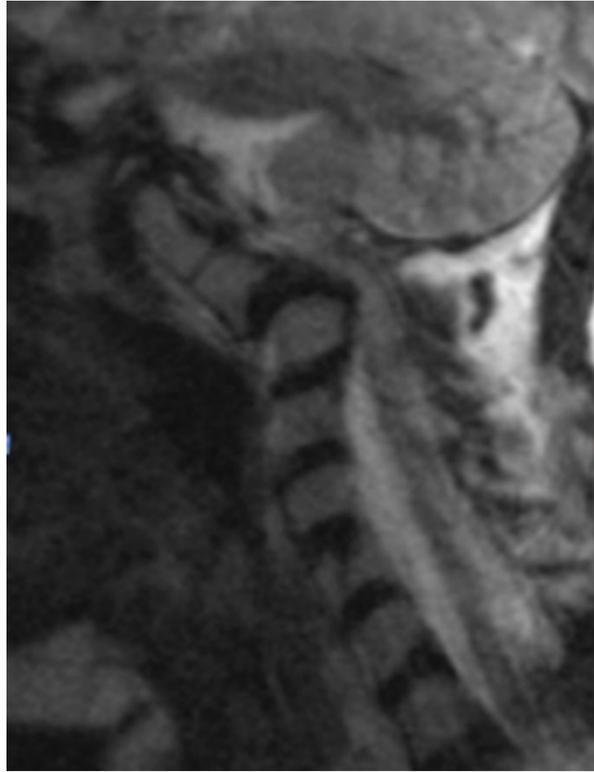
# Results

## Cervical Spine



6 years old

C2-C3 kyphosis and subluxation



10 years old



# Results

## Cervical Spine

### Treatment

- 7 patients fused for cervical spine instability
- Mean age at surgery 5.3 years, range 1 – 10 years
- Postoperative complications
  - Pseudarthrosis in 3 patients
  - 7 reoperations (bone graft placement, implant replacement)



# Results

## Thoracolumbar Spine

- Scoliosis  $> 25^\circ$  present in 15 of 39 patients (38%)
- 7 patients had primary thoracic curves
  - Magnitude  $52^\circ \pm 24^\circ$
  - Apex T9  $\pm 3$
- 8 patients had primary TL/L curves
  - Magnitude  $39^\circ \pm 11^\circ$
  - Apex L3  $\pm 1$



# Results

## Thoracolumbar Spine

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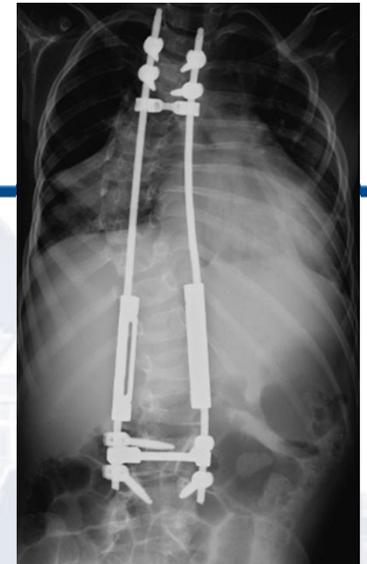
- Thoracic kyphosis  $> 45^\circ$  present in 9 of 39 patients (23%)
- Spondylolisthesis present in 11 of 39 patients (28%)
  - Grade  $2.5 \pm 1.1$
- Dural ectasia present in 18 of 30 patients (60%)

# Results

## Thoracolumbar Spine

### Treatment Scoliosis:

- 5 patients required surgery
- 4 patients had growing rods; 1 had definitive fusion
- 2 patients had occiput to sacral fusion
- 4 patients developed complications
  - Broken rods in 2 patients
  - Junctional kyphosis requiring extension of instrumentation in 1 patient
  - Tension pneumothorax in 1 patient
- 12 operations total



Immediate Postoperative



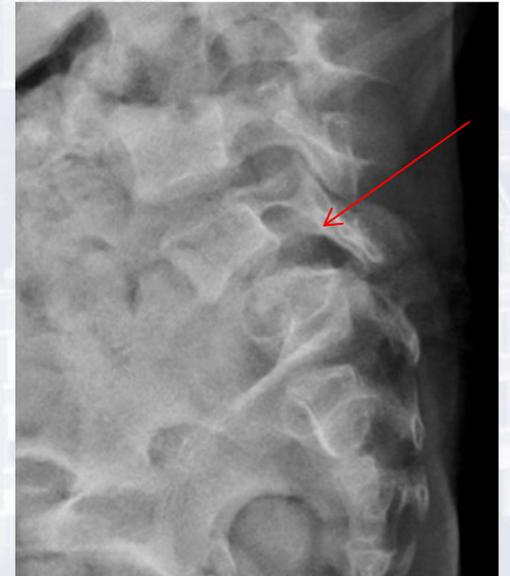
4 Year Postoperative

# Results

## Thoracolumbar Spine

### Treatment Spondylolisthesis:

- 6 patients required surgery
- 2 patient developed complications
  - Re-slip in 1 patient
  - Symptomatic implant requiring removal in 1 patient
- 8 operations total



# Limitations

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1. Incomplete records: Not all areas of the spine were imaged for every patient.
2. Sampling bias: The more severely involved patients may have been treated at our institution.

# Conclusions

- Cervical spine instability (subaxial), thoracic and thoracolumbar/lumbar scoliosis, thoracic kyphosis, spondylolisthesis, and dural ectasia are common in children with LDS.
- Spine deformity may require periodic re-screening of the entire spine.
- The prevalence of cervical and thoracolumbar spine abnormalities in this age group may be greater than previously reported in LDS patients of all ages.



# Thank you

