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High Incidence of Cervical Deformity and Instability Requires Surveillance in Loeys-Dietz Syndrome

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Background

Loeys-Dietz Syndrome (LDS)

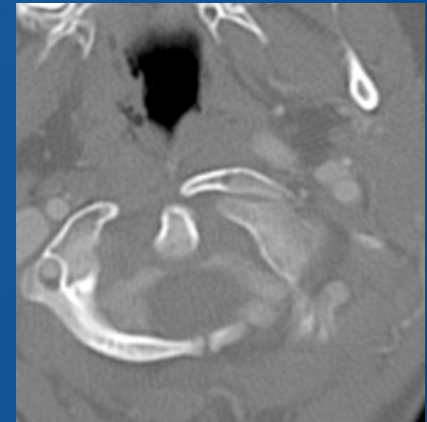
- Mutations in TGF- β R genes
- Characterized by
 - aneurysms
 - midline defects
 - skeletal deformities
- Objective: Study
 - 80 patients
 - Mean age 17.3
 - Mean follow up 6.5 years
 - describe
 - Incidence
 - Treatment & Complications



Results

C1 Hypoplasia

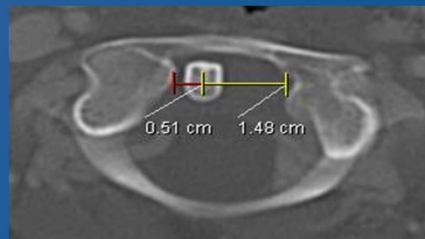
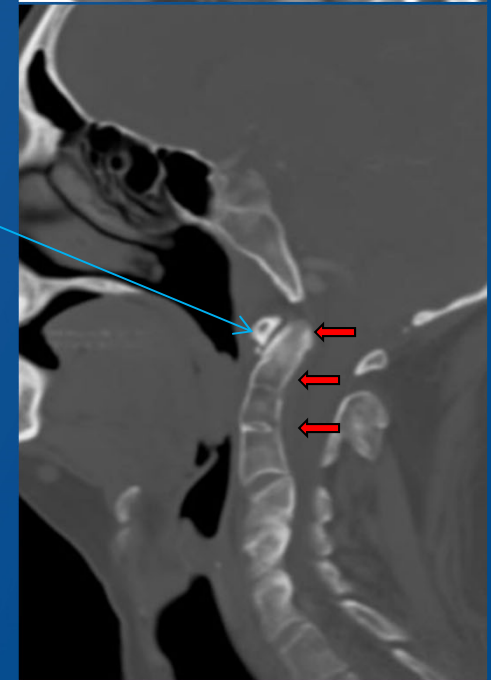
- Anterior arch defects more common than posterior arch defects
 - Anterior: 24%
 - Posterior: 16%



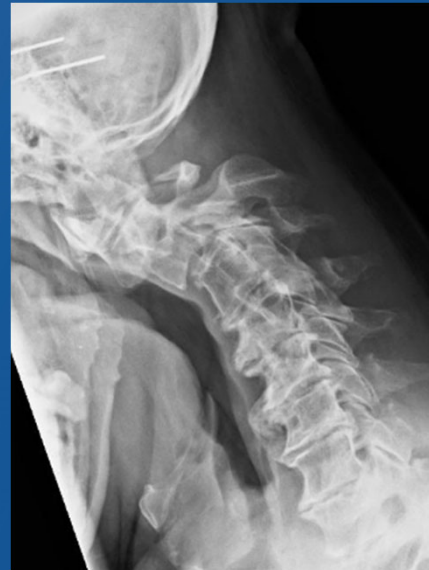
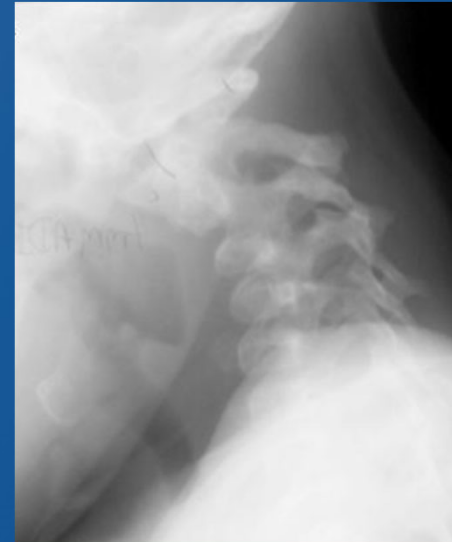
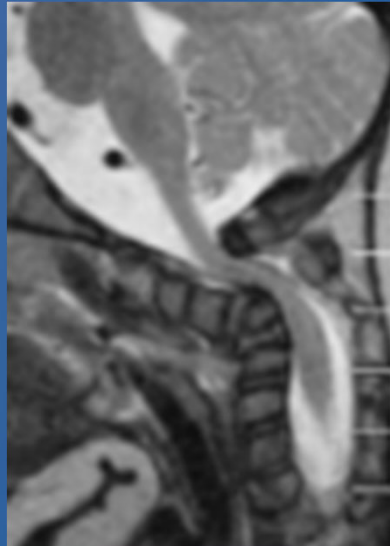
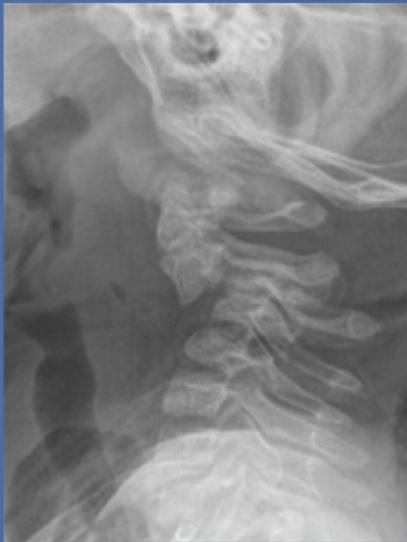
Results

C2 Deformity

- Elongation: 54%
- Dorsal angulation: 15%
- Spondylolysis: 5%
- Off center location: 21 patients (31%)



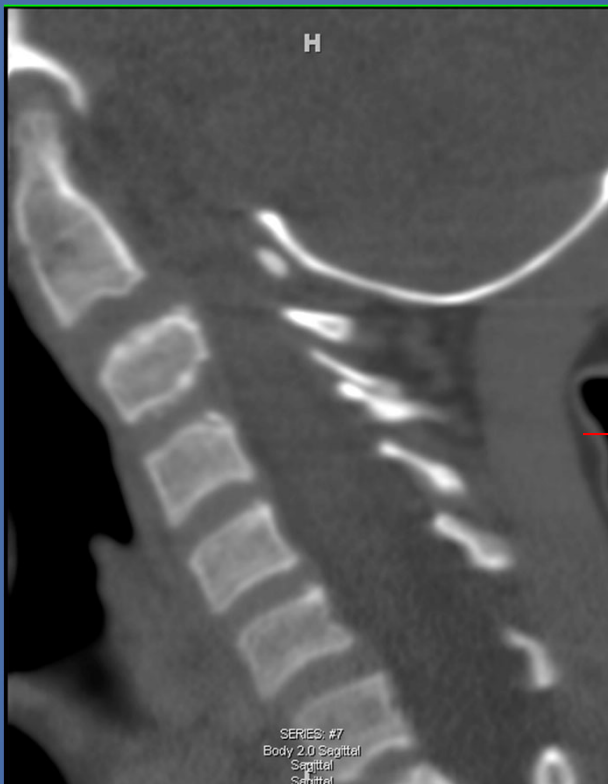
Sub-axial Deformity (9 pts) C3 Hypoplasia and instability



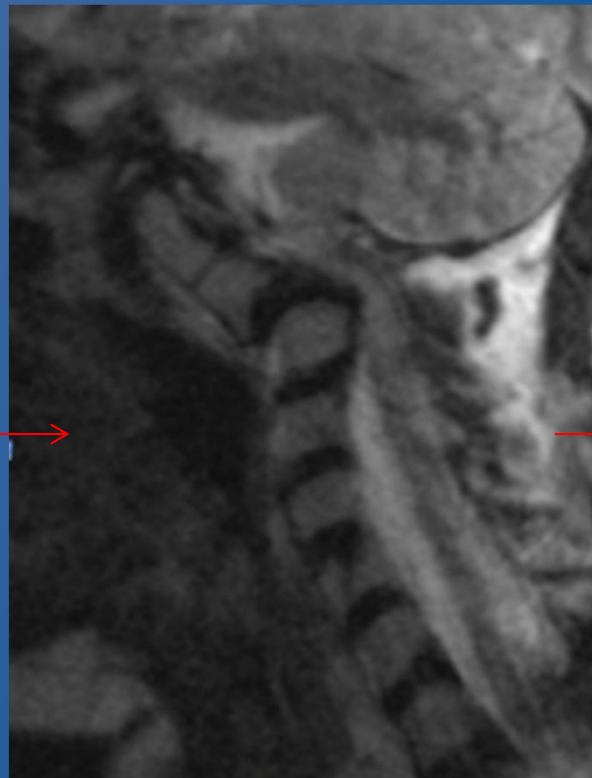
Subluxation in Late Childhood

-requires monitoring

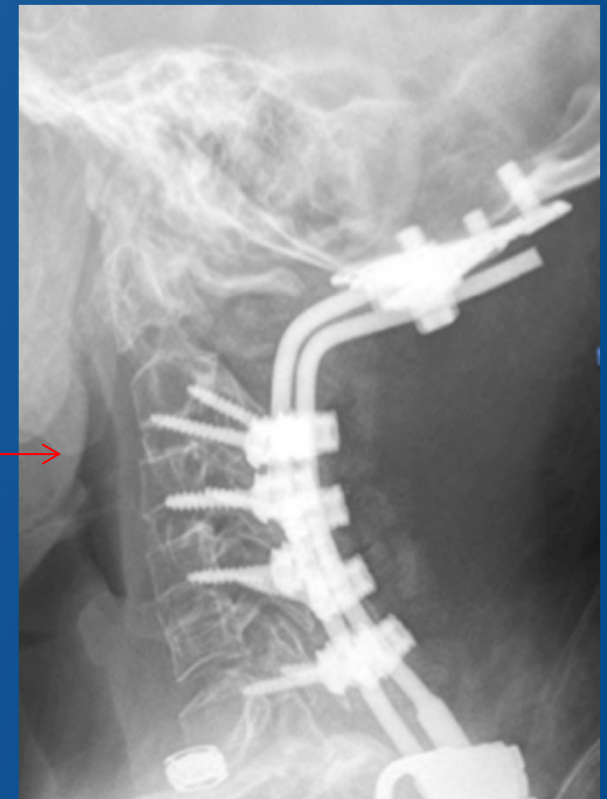
C2-C3 kyphosis
and subluxation



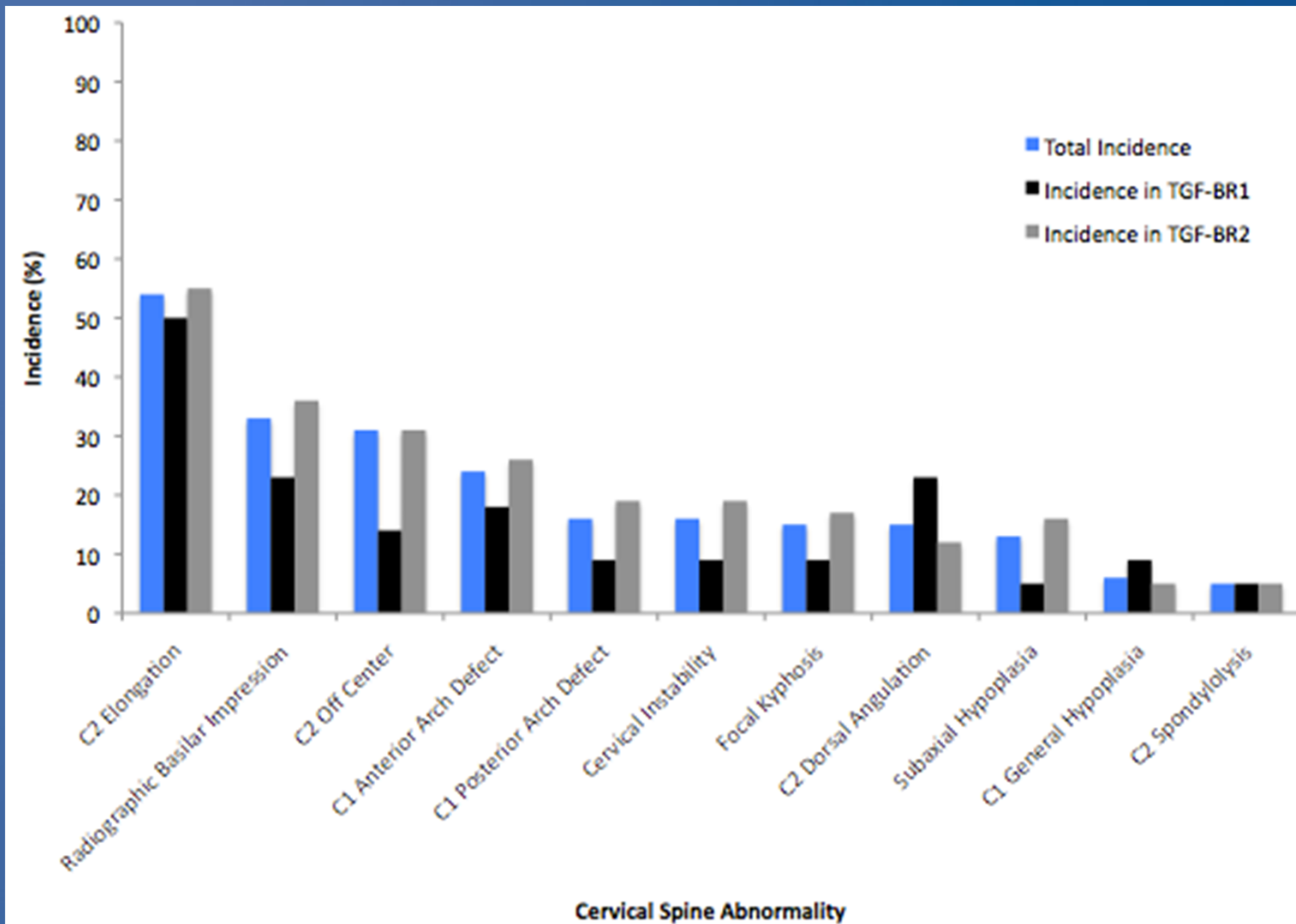
6 years



11 years



Type of TGF- β R Mutation NOT Associated with Cervical Abnormalities



Results

Treatment and Complications

- Spinal fusion for instability: 8 patients
 - Mean age 4.0 (range 3m-12y),
6 patients had surgery before age 3y
 - All had posterior, and 2 had anterior fusion
 - All fusions eventually involved occiput
 - All had neuro Sx / symptoms
- Complications: 7 patients, 13 reoperations
 - Pseudarthrosis: 5 (4 had failure of fixation)
 - Junctional kyphosis or instability: 4 patients



Conclusions

Cervical deformity & instability common in LDS

- C1-C3 most common
 - C1 arch defects, C2 spondylolysis, and C3 hypoplasia
- Most common is C3 hypoplasia with kyphosis and instability
- Recommend imaging & follow C-Spine
 - With growth

Thank you



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