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

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Loeys-Dietz Syndrome (LDS)

- Mutations in TGF-βR 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-βR 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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