

Age-Related Cervical Spine Deformities in Loeys-Dietz Syndrome- Evidence-Based Recommendations for Imaging

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INTRODUCTION:

Loeys-Dietz syndrome (LDS) is a genetic connective tissue disorder characterized by vascular and musculoskeletal malformations. Cervical spine abnormalities have been documented in LDS patients. We hypothesize that cervical deformities in patients with Loeys-Deitz Syndrome may become apparent after first imaging, so evidence-based screening recommendations are needed for monitoring.

METHODS:

We conducted a retrospective review of 92 patients diagnosed with LDS before the age of 18 who had available cervical spine imaging. Cervical images were evaluated for atlantoaxial or subaxial instability (>4 mm translation), focal kyphosis, stenosis (>15 degrees), and other cervical spine anomalies. Patient age at the time of each finding was recorded. Additionally, we examined the impact of thoracolumbar spinal fixation on the subsequent development of cervical spine pathology. All imaging findings were independently confirmed by a board-certified pediatric orthopedic surgeon. Descriptive statistics were used to characterize the frequency, distribution, and time interval from normal imaging to the onset of cervical pathology.

RESULTS:

Cervical spine abnormalities were detected in 45 of 92 patients (49%) despite normal initial imaging. Among patients with initially normal imaging who later developed findings (n=25), subaxial instability was the most common (44%, n=11), followed by focal kyphosis (40%, n=10), atlantoaxial instability (28%, n=7), and defects involving the atlas, axis, or odontoid (16%, n=4). The average time from a normal image to detection of pathology was 3.8 year. Patients who underwent thoracolumbar (TL) fixation were noted to develop secondary cervical deformities, with the most common being focal kyphosis.

DISCUSSION AND CONCLUSION:

Nearly half of LDS patients developed cervical spine abnormalities despite normal baseline imaging, with cervical instability being the most frequent finding. Repeat cervical imaging is recommended at least every three years. Patients undergoing thoracolumbar fixation should be closely monitored for subsequent cervical changes, as postoperative cervical deformities may develop due to the underlying connective tissue laxity in LDS. Given the potential for neurological compromise, early identification and intervention are critical to prevent serious complications.

Figure 1: Focal Kyphosis Requiring Cervical Fusion



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Figure 3: Cervical Kyphosis developed after T2-L4 PSF in 2020



Figure 2: Worsening Sub-axial Instability

