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Rapidly progressive scoliosis in a patient with Marshall-Smith Syndrome

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Rapidly Progressive Scoliosis in the Setting of Atlantoaxial Rotary Instability and Marshall-Smith Syndrome

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Abstract

Case: Marshall-Smith Syndrome is a rare genetic condition due to mutations in the NFIX gene resulting in a multitude of congenital abnormalities, including atlantoaxial instability and ligamentous laxity. This case report describes a patient with Marshall-Smith Syndrome who developed atlantoaxial rotary subluxation and subsequent rapidly progressive scoliosis after occiput-C2 fusion. She was treated with 4 weeks of halo traction, followed by an occiput-T3 posterior spinal fusion, segmental fixation, and magnetic growth rods from T3-L4. The thoracic curve was reduced from 125 to 34 degrees.

Conclusion: The presence of atlantoaxial rotary subluxation in the setting of generalized ligamentous laxity likely contributed to the rapid progression of scoliosis in this patient. Care should be taken to assess the rotatory stability of the atlantoaxial joint in patients with Marshall-Smith Syndrome and atlantoaxial instability. Unrecognized atlantoaxial rotatory instability in these patients can lead to serious consequences and the need for extensive surgical intervention.

Clinical and Surgical Course

- Female with genetically confirmed Marshall-Smith syndrome. She had global weakness as well as motor and cognitive developmental delays.
- Extensive surgical history, including tracheostomy, mandibular distraction for micrognathia, and ventriculoperitoneal shunt for hydrocephalus.
- Occiput-C2 fusion performed at 4 years of age. Preoperative and postoperative radiographs shown in figures 1 and 2, respectively.
- Developed torticollis and head tilt 1 year postoperatively. Referred for management of progressive kyphosis and scoliosis.
- Initial treatment consisted of cervical thoracic lumbar sacral orthosis with chin pad to slow deformity progression.
- Over 20 months, clear progression of rotating thoracic kyphosing scoliosis and lumbar curve, with restrictive pulmonary changes requiring supplementary oxygen therapy.
- Treated with halo traction at 50% body weight prior to planned occipital cervical to thoracic fusion and growth rods. Magnitude of curve reduced to 64 degrees in thoracic segment and 33 degrees in lumbar segment.
- After 4 weeks in halo traction, occiput-T3 posterior spinal fusion, segmental fixation, and magnetic growth rods from T3-L4 was performed. Thoracic curve was reduced to 34 degrees.

Discussion

- Craniocervical junction instability on flexion-extension radiographs has been reported in patients with Marshall-Smith Syndrome, but atlantoaxial rotary subluxation has not been described in this patient population.
- Underlying rotatory instability was not diagnosed at the time of the initial occiput to cervical fusion as the preoperative CT showed no pathologic rotation. The initial postoperative course was challenged by the patient writhing and thrashing, and rotation may have occurred in this time. This likely led to persistent rotatory subluxation and head tilt. The spinal deformity was not present initially but developed over the ensuing year postoperatively.
- Canale et al. reported on a series of otherwise healthy patients with congenital muscular torticollis. They found that at an average of 18.9 year follow-up patients with persistent head tilt developed a mild compensatory scoliosis.
- Adam et al. described soft tissue and general joint hyperextensibility in patients with Marshall-Smith Syndrome. The persistent head tilt and general ligamentous laxity in this patient may have ultimately led to the rapidly progressive scoliosis.
- To restore proper coronal and sagittal alignment, an occiput to L4 posterior construct was performed. Multiple studies have demonstrated the effectiveness of magnetically controlled growing rods for the treatment of early onset scoliosis. Additionally, there is significantly reduced morbidity and complication rates with magnetically controlled growing rod compared to traditional growing rod constructs.

Radiographs

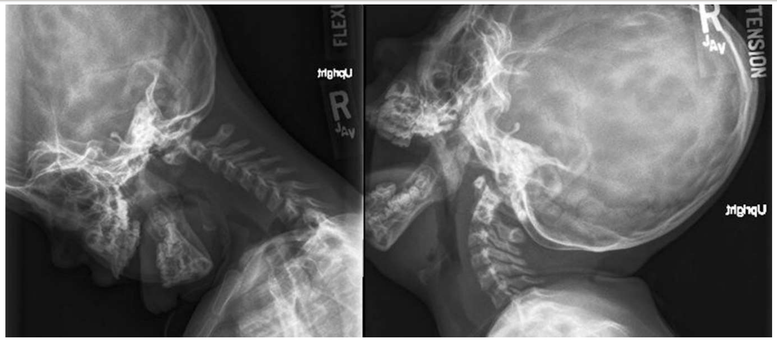

Figure 1. Flexion and extension lateral radiographs of the cervical spine demonstrating an atlanto-dens interval of 7.6 mm with flexion.

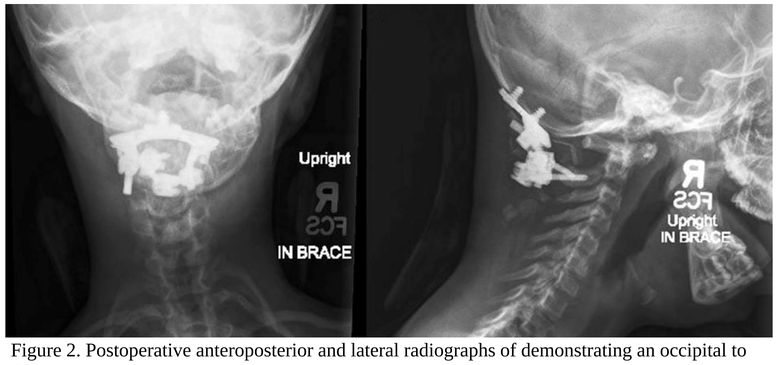

Figure 2. Postoperative anteroposterior and lateral radiographs of demonstrating an occipital to C2 fusion.

Figure 3. Posterior-anterior and lateral radiographs of the spine showing a rotating thoracic kyphosing scoliosis of 125 degrees and a lumbar curve of 64 degrees with significant head tilt.

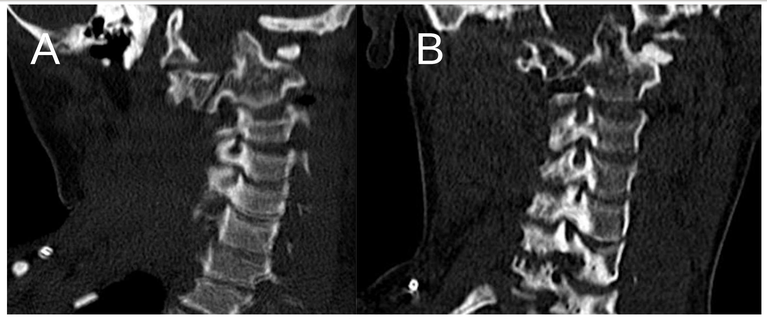

Figure 4. Computed tomography images of the cervical spine showing (A) progressive atlantoaxial rotary instability and (B) erosion of the right C6-C7 facet joint.

Figure 5. Postoperative posterior-anterior and lateral radiographs after occiput to T3 posterior spinal fusion, segmental fixation, and magnetically controlled growing rods from T3-L4.

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