

Successful cranio-cervical fusion in a patient with Down syndrome

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ABSTRACT

We present a patient with Down syndrome with neck pain and severe cervical myelopathy. Imaging revealed occipito-atlantal and atlanto-axial instability with severe spinal cord compression. There are no standardized radiological or clinical guidelines to aid in managing this unique subset of patients. We demonstrate a successful occiput–C3 internal fixation and fusion without complication. Due to the largely unknown natural history and ongoing management difficulties in this population, we demonstrate a case that may aid future decision making for this specialized field. We also discuss an approach to reduce this deformity, which, to our knowledge, has not been published before.

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1. Introduction

Down syndrome is the most common human chromosomal abnormality, and has been associated with myelopathy from craniovertebral instability. It is felt that symptomatic patients should be offered surgical stabilization. This report demonstrates a successful surgical approach in a young female with myelopathy and instability.

2. Case report

A 14-year-old female patient with Down syndrome presented with a 2-year history of neck pain; progressive ataxia; limb weakness and in coordination. Clinically she had minimal neck rotation, was hyperreflexic with sustained clonus, and had mild limb weakness. Lateral cervical radiographs showed occipito-atlantal and atlanto-axial subluxation. Subsequent investigations confirmed subluxation of C1/2 with an atlanto-dens interval (ADI) of 10 mm and vertical subluxation of the odontoid process, and subluxation of occiput/C1, with foramen magnum and spinal canal narrowing (Fig. 1). MRI confirmed myelomalacia.

The patient underwent a 7-hour occiput–C3 internal fixation and fusion. Through a suboccipital approach, internal fixation was achieved with C1 lateral mass, C2 pars and C3 lateral mass screws, including an occipital plate and rod with iliac crest bone graft bilaterally. Subtraction osteotomy of the C2 pars was performed to reduce the lateral mass of C1 onto the superior facet of C2 (Fig. 2). An iliac crest bone graft with tricalcium phosphate was used. Immobilization in a Miami-J orthosis for 3 months was used post-operatively. CT scans showed solid fusion at 6 months (Fig. 3).

3. Discussion

The etiology of craniovertebral abnormalities is diverse and can be a result of acquired, traumatic or inflammatory causes. It is associated with achondroplasia, mucopolysaccharidosis, and Klippel Feil syndrome¹ Craniovertebral abnormality has been associated with Down syndrome since first described in 1961.² The natural history is poorly understood, but the consensus is that the instability is chronic and progressive.³ The variety of

abnormalities includes atlanto-axial subluxation, occipito-atlantal instability and os odontoideum. Neurological sequelae, through sudden or progressive neurological deficit, has raised interest since the special olympics ruling for mandatory cervical spine imaging in Down syndrome athletes participating in “high risk” sporting activities.^{4,5}

There is no standardized link between radiological instability and patient symptomatology, and longitudinal studies have been unable to predict patient outcomes.^{5,6} Studies have shown atlanto-axial instability (AAI) present in 10% to 30% of this population, yet only 1% to 2% of this population show signs of neurological compromise.⁵ Tredwell et al.⁷ report occipito-cervical hypermobility in 60% of patients with Down syndrome; however, no patient showed signs of neurological compromise.

Early studies into craniovertebral instability assessed AAI, with ligamentous laxity and odontoid process abnormalities. Konttinen⁸ showed that patients with anterior atlanto-axial subluxation from transverse ligament rupture displayed mild myelopathy only, whereas alar ligament disruption leads to disruption of C1/2 facet joints and vertical subluxation of the odontoid process causing progressive neurological deficit.

Recent studies highlight abnormal bony anatomy in patients with Down syndrome as a factor in progressive myelopathy. Taggard et al.,⁹ and more recently Matsunaga et al.,¹⁰ demonstrated C1 hypoplasia as a factor in progressive cervical myelopathy in these patients. Furthermore, in this study the difference in ADI was not statistically significant between patients with Down syndrome and controls, whereas hypoplasia of C1 was, suggesting bony abnormalities of the craniovertebral skeleton in patients with Down syndrome are more significant risk factors for progressive neurological deficit than appreciated previously.

Studies published on outcomes of stabilization are few and, in earlier years, advocated extreme caution. Doyle et al.⁴ reported a 73% complication rate in 15 patients with Down syndrome who underwent posterior cervical fusion for AAI, with 40% requiring reoperation. They achieved fusion in 80%, but only a 20% symptomatic improvement. These findings were supported by Segal et al., who found a 100% complication rate with surgery.¹¹

The evolution of cervical instrumentation has improved surgical outcomes and more recent reviews show fusion rates as high as 58% to 95%.³ In addition, as in our patient, surgical stabilisation was achieved without fixed external immobilisation.

Our patient highlights the successful treatment of a young female patient with severe progressive cervical myelopathy due to

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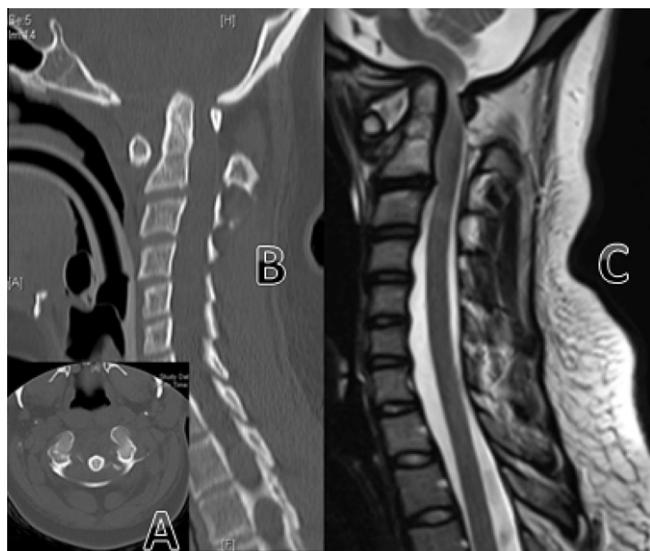


Fig. 1. Pre-operative images of a patient with Down syndrome: (A) axial CT scan at the level of the occiput/C1 demonstrating the odontoid peg migrating into the foramen magnum; (B) sagittal CT scan demonstrating C1 subluxation on C2 with migration of the odontoid peg into the brainstem; and (C) sagittal T2-weighted MRI demonstrating anterior subluxation of the occiput on C1 by 5 mm, anterior subluxation of C1 on C2 by 11 mm, and severe canal stenosis.

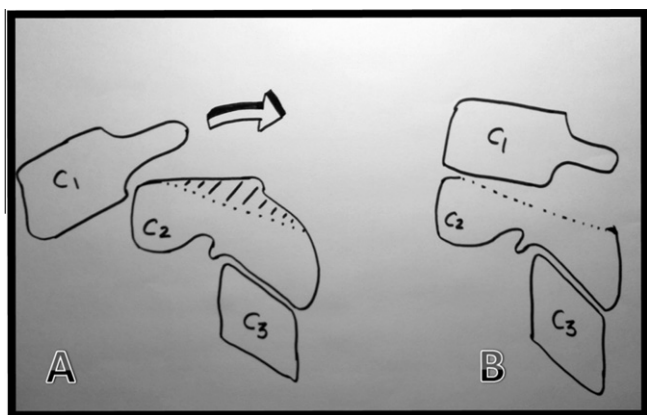


Fig. 2. Diagram representing a C2 subtraction osteotomy. The technique involves (A) an osteotomy of the superior aspect of the C2 pars and superior articular facet; and (B) the subtraction technique provides space to perform a reduction manoeuvre of the C1 lateral mass posteriorly to realign the C1/2 articulation.

significant ligamentous disruption coupled with bony abnormality.

4. Conclusion

Patients with Down syndrome with symptomatic cervical myelopathy from craniocervical instability pose a difficult subgroup of patients to manage surgically. Our patient highlights successful posterior occipito-cervical fusion, without complication or fixed post-operative external immobilization. Secondary to issues of compliance, safety and operative risk associated with this subset



Fig. 3. Post-operative images of a patient with Down syndrome: (A) mid-sagittal CT scans demonstrating occipital plate and reduction of occipital-C1 subluxation; (B) para-sagittal CT scans demonstrating lateral mass screw placement in C1–3; (C) T2-weighted MRI demonstrating improvement in the degree of canal stenosis at the occipital-cervical junction; and (D) para-sagittal CT scan at 6 months demonstrating solid fusion.

of patients, and a poor understanding of the natural history of the pathology, we advocate careful consideration of each case, and ongoing contributions of case series to the literature.

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