Original Article

Surgical strategies in the management of atlantoaxial dislocation in Down syndrome

ABSTRACT

Aims: To study the clinicoradiological features and treatment outcomes of atlantoaxial dislocation (AAD) in Down syndrome.

Settings and Design: Retrospective case series.

Subjects and Methods: A retrospective chart and radiology review of 9 Down syndrome patients with AAD managed at our center from 2007 to 2018.

Statistical Analysis Used: Chi-squared/Fisher's exact test.

Results: There were 4 males and 5 females ($n = 9$). The median age was 14 years (interquartile range [IQR]: 7–15.5). 77.7% (7/9) of patients had severe spasticity (Nurick Grades 4 and 5). The median duration of symptoms was 9 months (IQR: 5–39). The AAD was reducible in all ($n = 9$) cases. Eight (88.8%) patients had os odontoideum. The mean atlantodental interval (ADI) was 8.5 mm (±2.9). T2W cord hyperintensity was seen in 66.6% (6/9). Posterior C1–2 transarticular fixation was done in 8 and occipitocervical fusion in 1 patient. Follow-up of more than 6 months (7–57 months) was available in 8/9 (88.9%) patients. There was a significant improvement in spasticity ($n = 8$, mean Nurick Grade 1.7 (±1.1), $P = 0.003$). Follow-up radiographs ($n = 8$) showed good reduction and fusion. A preoperative bedbound patient with poor respiratory reserve expired at 10 months following surgery. There were no other complications.

Conclusions: Posterior surgical approach for AAD in Down syndrome resulted in good alignment and fusion, with excellent clinical improvement. Patients with elevated PCO_2 are poor surgical candidates and require home ventilation facility.

Keywords: Atlantoaxial dislocation, Down syndrome, os odontoideum

INTRODUCTION

Instability at the craniovertebral junction occurs in one-third of patients with Down syndrome due to laxity of the ligaments, decreased muscle tone, odontoid hypoplasia, or os odontoideum.[1] This problem was highlighted in 1983 by the Special Olympics Committee which prohibited such individuals from participating in high-risk sporting events.[2,3] Atlantoaxial instability occurs in children and adolescents with Down syndrome, but the majority of these patients are asymptomatic. This instability can be diagnosed on routine lateral cervical spine radiographs by measuring the atlantodental interval (ADI).[4] Data regarding outcomes for the management of these patients are limited. Asymptomatic patients without os odontoideum can be followed up and managed without surgery. In the present study, we have reported our experience in managing the symptomatic patients over the last 11 years and have suggested a practical management algorithm to aid clinical decision-making.

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SUBJECTS AND METHODS

This retrospective study was performed on a historical cohort of nine Down syndrome patients with atlantoaxial dislocation (AAD) managed in a single neurosurgical unit at our center between 2007 and 2018. The demographic data, presenting symptoms, duration of symptoms, functional status as determined by Nurick grade, details of intervention, and outcomes were obtained from the hospital electronic database. The clinical and radiological profile, management strategies, and outcomes of intervention were studied.

The primary outcome measure was the change in functional status measured by the Nurick grade at follow-up. All patients had cervical spine X-ray anteroposterior (AP) views, lateral neutral and extension views, computed tomography (CT), and magnetic resonance imaging (MRI) of the cervical spine (including the craniovertebral junction) preoperatively. Plain cervical spine X-rays (open-mouth AP, lateral flexion, and extension views) and X-ray tomograms were done postoperatively and at follow-up. Postoperative CT and MRI scan of the cervical spine were also done in the last four patients. These images were reviewed on our Picture Archiving and Communication System (GE-PACS version 3.0). Radiological parameters studied included the measurement of ADI on lateral neutral cervical spine X-rays, reduction of ADI on extension radiographs, presence of an os odontoideum, and cord T2W hyperintensities at the craniovertebral junction. The presence of basilar invagination (BI) and associated bony anomalies such as assimilation of the atlas and fused subaxial vertebrae was also documented.[5]

Statistical methods

The data collected were entered into a spreadsheet and statistical analysis was performed using SPSS version 21.0 (IBM, Bengaluru, Karnataka, India). Descriptive statistics were reported using mean ± standard deviation for continuous variables and median (interquartile range [IQR]) for skewed variables. Categorical variables were reported as frequency and percentage. Association of variables was reported using Chi-squared/Fisher's exact test. Comparison of means was reported using two independent samples *t*-test. $P < 0.05$ was considered statistically significant.

RESULTS

Table 1 summarizes the preoperative clinicoradiological data of our patients.

The study population consisted of four males and five females. The median age group was 14 years (IQR: 7–15.5 years). Myelopathy was the presenting complaint in 8 (88.8%) patients and the median symptom duration was 9 months (IQR: 5–39 months). One (11.1%) patient presented with neck pain. Additional clinical findings noted were torticollis in 2 (22.2%), urinary incontinence in 3 (33.3%), and breathing difficulty in 1 (11.1%) patient. Arterial blood gas analysis was done in all patients who had severe spasticity at presentation (Nurick Grades 4 and 5, $n = 7$). One of these seven patients had breathing difficulty at presentation and his PCO_2 value was 51.2 mmHg. The remaining six patients had normal PCO_2 values and did not have any difficulty breathing.

Table 1: Clinical characteristics of Down syndrome patients with atlantoaxial dislocation

Variable	Number of patients (%) ($n=9$)
Gender	
Male	4 (44.4)
Female	5 (55.5)
Clinical characteristics	
Presenting complaints	
Neck pain	1 (11.1)
Myelopathy	8 (88.8)
Additional findings	
Torticollis	2 (22.2)
Urinary incontinence	3 (33.3)
Breathing difficulty	1 (11.1)
Preoperative Nurick grade	
Grade 0	1 (11.1)
Grade 2	1 (11.1)
Grade 4	2 (22.2)
Grade 5	5 (55.5)

Preoperative neurological function

Seven (77.7%) of the nine patients had severe spasticity at presentation and were Nurick Grades 4 or 5. One (11.1%) patient was Nurick Grade 2 at presentation, and 1 (11.1%) patient presented with only neck pain.

Radiology

The mean ADI measured on lateral neutral cervical spine X-ray was 8.5 mm (±2.9 mm), and reduction in the ADI could be demonstrated on preoperative extension lateral cervical spine X-rays, in all patients [Figures 1 and 2]. Os odontoideum was present in 8/9 (88.8%) patients [Figure 3] and 1 patient had an ossiculum terminale [Figure 1]. T2-weighted cord hyperintensities were seen in 66.6% (6/9) of our patients [Figure 4]. None of our patients had assimilation of atlas, BI, or fused subaxial vertebrae. These findings are summarized in Table 2.

Management

All patients underwent posterior stabilization and fusion. C1–2 transarticular fixation with C1–2 cable and bone graft placement (modified Gallie's fusion)[6] was done in 7 patients.

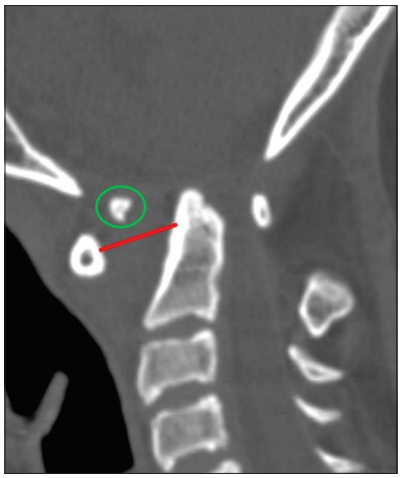

Figure 1: Midsagittal slice of the computed tomography scan of the craniovertebral junction showing widening of the atlantodental interval (solid red line) with narrowing of the spinal canal, and ossiculum terminale (green circle)

Transarticular fixation with bone graft placement without modified Gallie's fusion was done in 1 patient, and the first patient in our series underwent occipitocervical fusion (OCF) with titanium loop and sublaminar wires, followed by halo vest fixation. In 7 cases, the transarticular screws were placed bilaterally, and in 1 case, transarticular screws were placed only on one side (left side). None of our patients were put on cervical traction preoperatively.

Operative technique

The patient was turned prone on a Philadelphia collar and the head was fixed on a Mayfield® (Integra life Sciences) three-pin head holder. The AAD was reduced as far as possible by elevating the occiput in military tuck position, under fluoroscopic guidance. Further reduction was achieved by palpating the C2 spinous process and pushing it anteriorly. Midline upper cervical incision was marked, along with two incisions about one finger breadth away from the midline at C7/T1 level. After making the skin incisions and deepening it to divide the deep fascia, the soft tissues were reflected off the C1–C2 posterior elements in a subperiosteal manner. Care was taken to dissect along the C2 pars "roof" toward the C1/2 lateral mass joint. The venous plexus over the C1/2 joint was carefully coagulated and divided and the C1–2 joint capsule opened into, staying inferior to the C2 root, which was not divided. The joint cartilage was then curetted, and morselized pieces of iliac crest autograft were placed inside the joint. A pilot hole was drilled at the inferior aspect of the C2 pars about 2.5 mm above and lateral to the medial border of the C2–3 facet joint. Separate stab incisions were made at C7-T1 level bilaterally, through which the Universal Cannulated Screw System (UCSS®, Medtronic Sofamor Danek USA, Inc) was introduced.

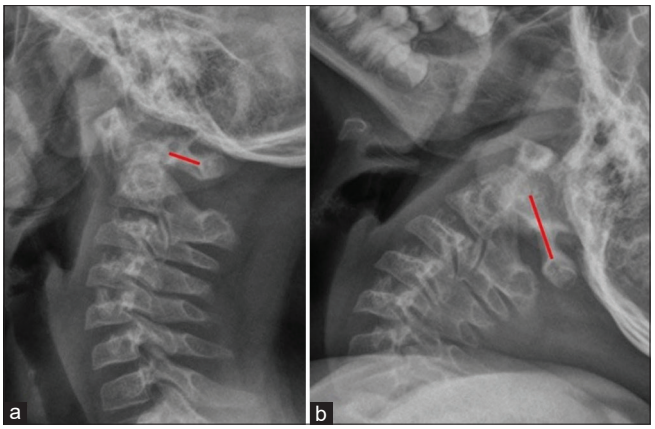

Figure 2: Dynamic lateral cervical spine radiographs in neutral (a) and extension (b) views, showing reduction of the atlantoaxial dislocation on extension (b), with a corresponding increase in the diameter of the spinal canal as denoted by the solid red line

Table 2: Radiological features of Down syndrome patients with atlantoaxial dislocation

Parameter	Number of patients (%) (n=9)
Reducible AAD	9 (100)
Os odontoideum	8 (88.8)
Ossiculum terminale	1 (11.1)
T2 weighted cord hyperintensities	6 (66.6)

AAD - Atlantoaxial dislocation

A "k" wire was placed in the pilot hole, and drilled through the C2 pars, across the C1/2 facet joint into the C1 lateral mass on one side initially (usually the left side). A cannulated drill bit was then used to drill over the k wire and this was followed by a cannulated tap. A measured cannulated screw was then placed over the k wire, and the k wire was removed after making sure that the fit was good. All these steps were under fluoroscopic guidance. These steps were then carried out on the other side, following which the C1/C2 complex was checked and seen to move as a single unit, with video fluoroscopy.

Types of screws used for the C1/2 transarticular fixation

4 mm dia. screws (UCSS® Medtronic Sofamor Danek USA, Inc) were used in 3 patients bilaterally, 3.5 mm dia. screws (Corridor® Fixation System, Globus Medical, Inc., Audubon, PA, USA) were used bilaterally in 2 and unilaterally in 1 patient, and 1 patient had both 4 mm UCSS® and 3.5 mm Corridor® screws placed one on each side. In one patient, we used 3 mm dia. screws from a small fragment set borrowed from our hand surgery orthopedic colleagues [Figure 4].

Modified Gallie's fusion

Atlas cable® (Medtronic Sofamor Danek USA, Inc) was passed under the C1 posterior arch and then over the C2 spinous process. A shaped piece of iliac crest/rib autograft was

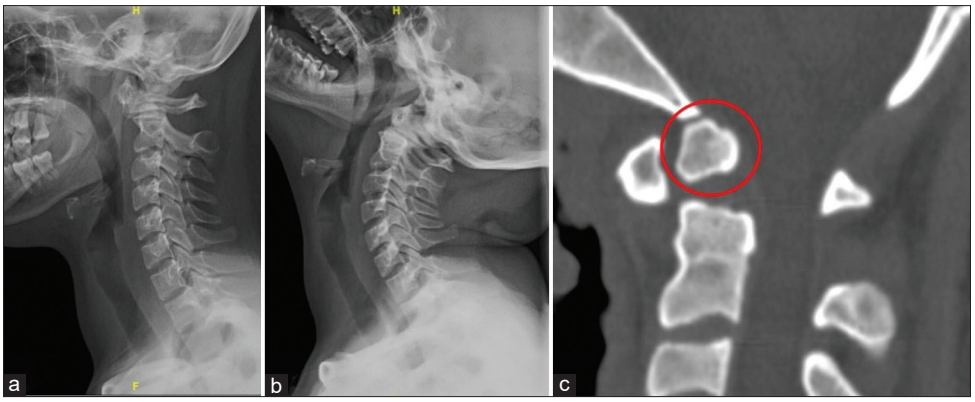

Figure 3: Radiographs of cervical spine in neutral (a) and extension (b), demonstrating the reducible atlantoaxial dislocation. Midsagittal computed tomography scan (c) of the craniovertebral junction showing an os odontoideum (red circle)

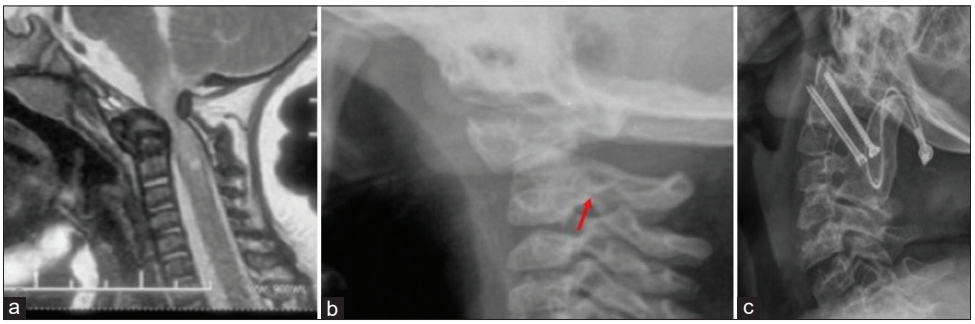

Figure 4: Midsagittal T2W magnetic resonance imaging showing severe compression with T2W cord changes (a). Lateral cervical spine X-ray (b) showing the atlantoaxial dislocation (AAD) and narrow C2 pars (red arrow). Follow-up (57 months) lateral X-ray (c) showing the reduced AAD with good bony fusion

placed between C1 and C2 posterior elements, and the cable was passed over the graft and again under the C2 spinous process (modified Gallie's fusion). It was finally tightened, crimped, and cut. Smaller pieces of iliac crest bone graft were then placed around this construct.

The first patient in our series underwent excision of posterior arch of C1 and OCF (C0–C3) with titanium pins, and secured with sublaminar wires passed under C3 lamina, and through the suboccipital bone on either side of the midline. Rib graft was placed, and following wound closure, halo vest was applied postoperatively.

Bone graft used

We used rib autograft in one patient and iliac crest autograft in the remaining 8 cases, to facilitate bony fusion.

All patients were advised to use a Philadelphia collar/hard collar for 3 months postoperatively.

Clinical and radiological outcome

All patients who presented with myelopathy had improvement in spasticity in the postoperative period. Follow-up data were available in 8 of the 9 cases (88.8%) – one patient was lost to follow-up. The median follow-up duration (n = 8) was 41.5 months (IQR: 8.5–67 months). Nurick grade at follow-up (n = 8) showed a significant improvement in the functional Nurick grade (P = 0.003) [Table 3]. The patient with neck pain had good relief of the pain.

There was good bony fusion (defined as trabecular bone formation at C1-2 and/or absence of instability on dynamic lateral cervical spine X-rays) at follow-up in all 8 cases (100%) [Figure 5]. One patient underwent tracheostomy in the immediate postoperative period as he developed worsening respiratory distress following extubation. He was a 5-year-old child who had breathing difficulty and was bed-ridden (Nurick Grade 5) preoperatively. Arterial blood gas analysis done before surgery showed an elevated PCO_2 (51.2 mmHg). Postoperatively, he was discharged on a tracheostomy and there was some improvement in his spasticity (Nurick Grade 4) at 10 months of follow-up. He developed respiratory arrest and expired 10 months postoperatively. There were no other complications in our series.

DISCUSSION

Management of atlantoaxial instability in Down syndrome patients is one of the most challenging conditions to deal with

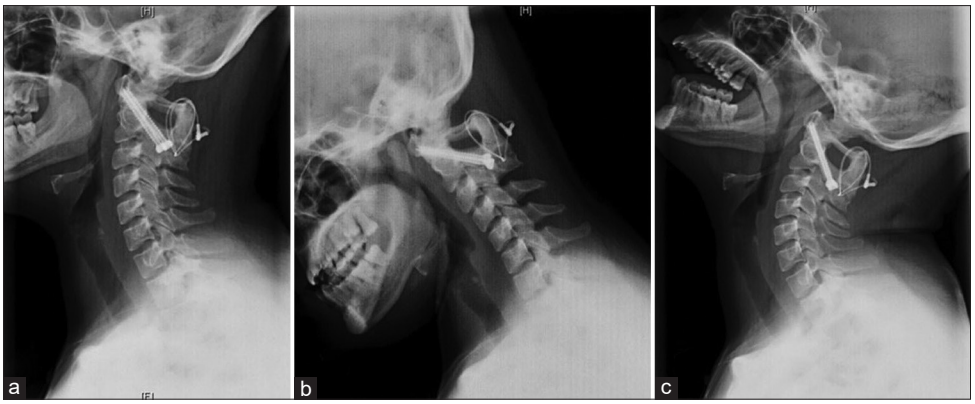

Figure 5: Follow-up cervical spine radiographs of the case shown earlier [Figure 3]. There is good trabecular bone formation at C1-2 (a) with reduction in atlantodental interval, and no instability in the flexion (b) and extension (c) images

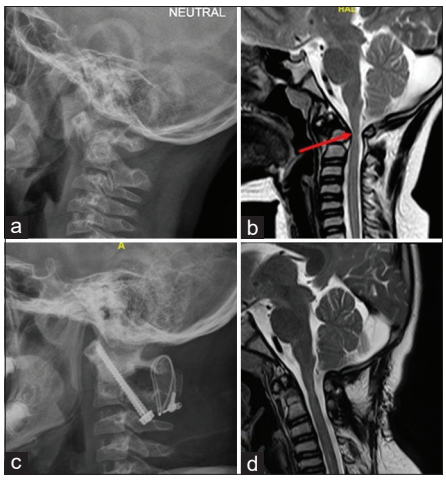

Figure 6: (a) Showing the atlantoaxial dislocation, which is reduced in the postoperative Xray (c). Preoperative midsagittal T2W MRI (b) showing anterior indentation of the cord (red arrow), which has resolved with good space for the spinal cord in the postoperative MRI (d)

both for the novice and experienced surgeon, especially in pediatric population. Our results clearly show that these patients can be managed with a single stage posterior approach alone.

AAD is reported in up to one-third of patients with Down syndrome and occurs due to laxity of the ligaments, decreased muscle tone, odontoid hypoplasia, or os odontoideum.[1] Children with Down syndrome have an abnormal shape of the occipitocervical articulation, which results in an increased anterior and lateral translation, thereby stressing the ligaments which eventually fail, resulting in occipitocervical and AAD.[7] Bony anomalies such as os odontoideum occur in approximately 6% of children with Down syndrome and contribute to atlantoaxial instability.[8] Eight patients in our series (n = 9) had an associated os odontoideum.

The natural history of ligamentous atlantoaxial instability without associated bony anomalies has been reported in Down syndrome with asymptomatic AAD. Burke *et al*.

Table 3: Postoperative follow-up (n=8) Nurick grade (mean Nurick grade: Preoperative 4±1.5, postoperative 1.75±1.16, P=0.003)

Preoperative Nurick grade	Postoperative Nurick grade						Total (n=8)
	0	1	2	3	4	5	
0	1	0	0	0	0	0	1
1	0	0	0	0	0	0	0
2	0	1	0	0	0	0	1
3	0	0	0	0	0	0	0
4	0	1	1	0	0	0	2
5	0	2	0	1	1	0	4

reported that 7/32 patients who demonstrated abnormal motion on initial evaluation had increase in ADI at a follow-up of 13 years.[4] Others have reported progressive decrease in the ADI and no change in neural canal width during a 5-year follow-up period in children with Down syndrome and AAD.[9,10] Therefore, it appears that ligamentous instability at the atlantoaxial joint is unlikely to progress to clinically relevant subluxation in the absence of bony anomalies in clinically asymptomatic Down syndrome patients.[11]

Management strategies and outcomes

Posterior stabilization is an effective treatment modality for the management of instability associated with reducible AAD.[12,13] We found that the AAD was reducible in all patients in our series. Posterior arthrodesis can be obtained by a variety of techniques such as OCF, C1–2 transarticular screw fixation (Magerl's), or Goel-Harm's C1–2 fixation.[14-18]

The use of contoured rods and wires is a simple cost-effective technique of achieving occipitocervical stabilization in patients with narrow pedicles/pars and ones who are at higher risks of vertebral artery injury.[19,20] The disadvantages of this technique included the need to extend the construct to longer levels to achieve the desired stabilization, sagittal plane deformity due to the crankshaft phenomenon, injury to the dura/spinal cord due to the passage of sublaminar

wires, and the need for prolonged immobilization with a halo vest postoperatively.[21,22] We had adopted this technique for the first patient in our series. With advances in spine instrumentation, sublaminar wiring techniques are no longer used, and more rigid methods of OCF are available, though the indications for this technique, i.e. including the occiput in the fusion, have reduced.

In a meta-analysis of twelve studies, Scollan *et al*. reported that the atlantoaxial transarticular screw (Magerl) technique and the screw-rod systems (Goel-Harm) technique were most frequently used posterior stabilization techniques in these patients.[23] The authors reported a number of complications related to posterior arthrodesis; the most common ones were bone related (39.6%). They suggested that the impaired skeletal maturity and bone mass leads to nonunion with subsequent loss of reduction. They found that nonunion occurred in 18.2% of cases, 14.2% of patients had fractures, and in 25% of cases, there was resorption of the bone graft, leading to poor fusion. The reoperation rate in their study was close to 34.9%.[23] In our series, we had good bony fusion in all our patients and none of the cases required reoperation.

The same authors also reported that patients with preoperative neurological deficits were at higher odds of neurological deterioration postoperatively,

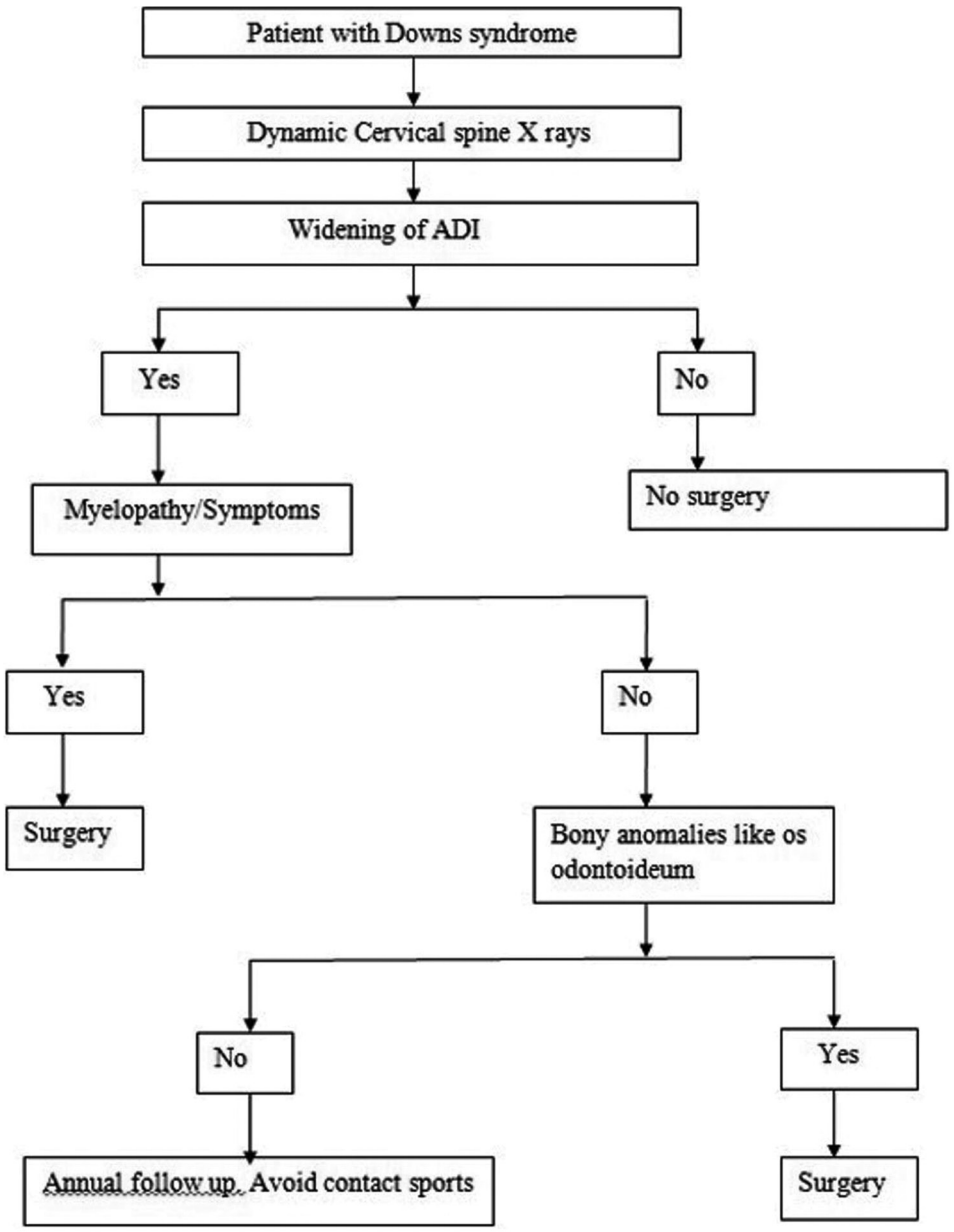

Figure 7: The proposed treatment algorithm. ADI: Atlantodental interval

compared to those without preoperative deficits.[23] This is in contrary to our findings as we found that the surgery results in decompression of the cord and stabilization of the AAD [Figure 6] and even patients with severe myelopathy benefit from surgery. Eight of the 9 patients in our series had spasticity at presentation, and 7 of these 8 were severely incapacitated due to the spasticity (Nurick Grades 4 or 5). One patient was lost to follow-up, but all of the remaining had good symptomatic improvement after surgery; there was improvement in the mean Nurick grade to 1.7 (±1.1), from the preoperative 4 (±1.5) ($P = 0.003$) [Table 3].

Strang and Katwa reported long-term follow-up of a 10-year-old child who underwent posterior fossa decompression for Arnold–Chiari malformation. This patient had severe central sleep apnea with a very high PCO_2 preoperatively.[24] The patient continued to have sleep apnea postoperatively and required intermittent positive pressure ventilation. The apnea was gradually improving at 7.5 years of follow up. We had one patient in our series who had elevated PCO_2 levels preoperatively; he underwent tracheostomy postoperatively and had been discharged on the same. Although there was improvement in his spasticity postoperatively, he developed respiratory arrest and expired 10 months postoperatively. Patients with elevated PCO_2 levels preoperatively need to be carefully evaluated and should be counseled regarding the need for long-term home ventilation.

Based on the observations in our cohort and review of literature, our strategy for the management of AAD in Down syndrome patients is summarized in Figure 7.

CONCLUSIONS

AAD associated with Down syndrome is usually reducible. Patients with associated bony anomalies such as os odontoideum and those with clinical worsening should be offered early surgery. Such patients can be managed with posterior only approaches. Majority of patients including those with poorer Nurick Grades 4 and 5 have a good outcome with surgery. Those with elevated PCO_2 levels preoperatively should be counseled about the need for long-term home ventilation.

Limitations of the study

This is a small case series of 9 cases with AAD in Down syndrome. Further studies with larger numbers would be useful to validate the treatment plan [Figure 7] for such patients.

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Nil.

Conflicts of interest

There are no conflicts of interest.

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