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CASE REPORTS

Sleep endoscopy-directed management of Arnold-Chiari malformation: a child with persistent obstructive sleep apnea

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Arnold-Chiari malformations are structural defects in the base of the skull and cerebellum, when part of the cerebellar tonsils herniates through the foramen magnum into the upper spinal canal, compressing against the brainstem. This anatomical defect can be asymptomatic but often presents with symptoms such as headaches, stridor, dysphagia, and nystagmus. It also presents with a variety of sleep-related breathing disorders such as snoring, obstructive sleep apnea, central sleep apnea, bradypnea, and sleep hypoventilation. Sometimes these conditions can coexist in one patient. Although obstructive sleep apnea can be a manifestation of Arnold-Chiari malformation, identifying causality and the site of obstruction in these children can be a diagnostic challenge. We review the case of a 14-month-old male who presented with noisy breathing and obstructive sleep apnea diagnosed on sleep study that was refractory to initial upper airway surgery. Although a brain computed tomography scan done in the emergency room for altered mental status revealed a type 1 Arnold-Chiari malformation, and a flexible awake laryngoscopy was normal, subsequent drug-induced sleep endoscopy was helpful in justifying surgical decompression of the Arnold-Chiari malformation that resulted in complete resolution of the obstructive sleep apnea.

Keywords: Arnold-Chiari malformation, drug-induced sleep endoscopy, hypoventilation, pediatric obstructive sleep apnea, stridor

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INTRODUCTION

Arnold-Chiari malformation (ACM) is caused by herniation (greater than 6 mm) of the cerebellar tonsils through the foramen magnum into the spinal column. Commonly, ACM can present with sleep-related breathing disorders such as snoring, central sleep apnea, bradypnea, hypoventilation, breath-holding spells, or obstructive sleep apnea (OSA).[1–3] The common anatomical cause of upper airway obstruction in otherwise healthy children is enlarged lymphoid tissue such as adenotonsillar hypertrophy. Therefore, adenotonsillectomy is considered as the first line of treatment of OSA in children.[4] Yet, it has been shown that in several studies and meta-analyses that this treatment strategy is not very effective in curing OSA in most children, and a thorough upper airway evaluation is essential to evaluate other sites of obstruction. Evaluation of these patients include other modalities such as laryngoscopy and drug-induced sleep endoscopy (DISE). These endoscopic techniques are often used to evaluate the upper airway for dynamic obstruction that may be amenable to additional surgical treatment.[5] Here we describe the case of a 14-month-old male who presented with symptoms of upper airway obstruction and who initially underwent adenoidectomy and supraglottoplasty. Following the surgery, the patient continued to have persistent obstructive breathing and based on the findings of DISE, a specific cause of OSA could be established and effectively treated. As per our institutional review board policy, a single case report is not considered human subject research and hence no institutional review board approval was necessary.

REPORT OF CASE

A 14-month-old male with a history of noisy breathing, cough, rhinorrhea, poor oral intake, and emesis presented to the emergency department. The mother stated the patient had two episodes of nonbilious nonbloody emesis, and in addition, the mother reported the patient's twin brother having similar symptoms days prior. There was no history of fever, diarrhea, or rash. Given the relatively uneventful course in the emergency department and improvement, he was discharged home with a diagnosis of viral upper respiratory infection and advised to follow up with his primary care physician. The following morning the patient was found to be lethargic and nonresponsive. The mother and the patient returned to the emergency department where he was intubated due to concerns for seizure activity and acute respiratory failure with a critically elevated pCO_2 (89 mm Hg). Noncontrast computed tomography of the head and brain magnetic resonance imaging (MRI) revealed subtle areas in the brain concerning for mild hypoxic injury as well as a ACM type 1.

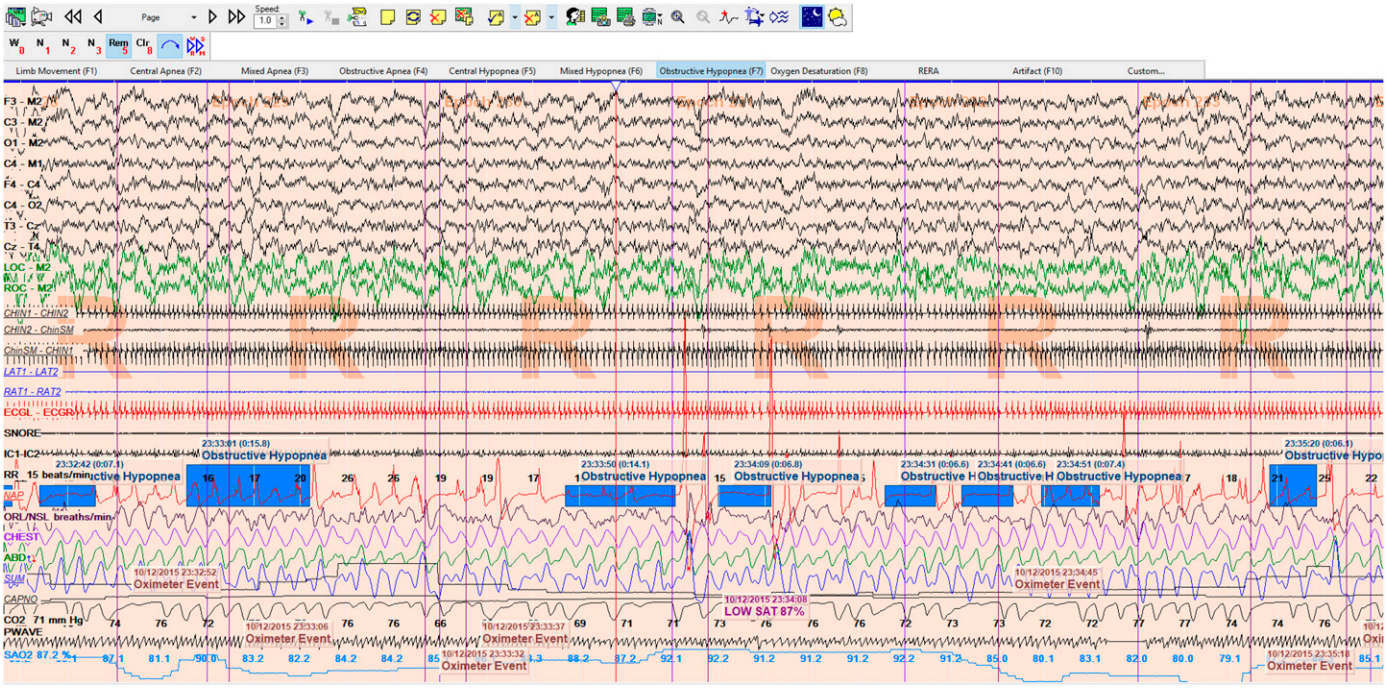
The patient was extubated on day 3 and postextubation, he exhibited persistent stridor, snoring, frequent desaturations, and

Table 1—Serial polysomnography results for initial preoperative and postoperative adenoidectomy and supraglottoplasty, and postoperative posterior fossa decompression studies.

	Preoperative PSG	Post Adenoidectomy and Supraglottoplasty (Split-Night Study) Baseline Portion	1-Month Post Decompression PSG	6-Month Post Decompression PSG
TST, minutes	402.0	240.0	487.5	420.0
REM sleep, minutes (%)	117.0 (29.1)	65.0 (27.0)	121.0 (24.8)	114.0 (27.1)
NREM sleep, minutes (%)	282.0 (70.1)	174.0 (73.0)	366.5 (75.2)	306.0 (72.9)
$ETCO_2$ > 50 torr, %TST	100.0	100.0	0.1	0.0
SaO_2 < 92%, %TST	10.5	2.5	13.0	0.0
SaO_2 nadir, %	74.1	80.0	68.8	96.5
OAHI, events/h	7.5	3.5	0.2	0.0
REM OAHI, events/h	12.3	11.0	1.0	0.0
NREM OAHI, events/h	5.7	0.7	0.0	0.0
Central sleep apnea, events/h	0.0	0.8	0.2	0.0

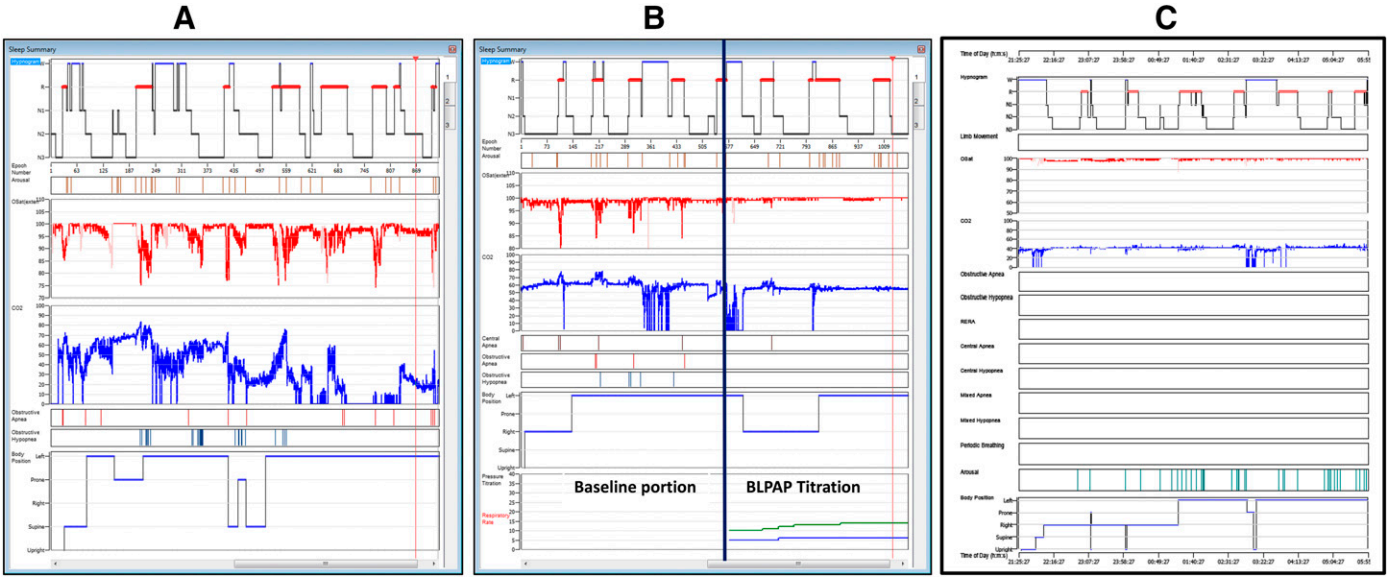
AHI = apnea-hypopnea index, $ETCO_2$ = end-tidal carbon dioxide, NREM = non-rapid eye movement, OAHI = obstructive apnea-hypopnea index, PSG = polysomnography, REM = rapid eye movement, SaO_2 = saturation of oxygen, TST = total sleep time.

Figure 1—A 3-minute epoch of polysomnography showing rapid eye movement sleep with frequent obstructive hypopneas associated with severe desaturation and hypercarbia.



increased work of breathing during sleep. A bedside flexible nasopharyngoscopy while the patient was awake noted markedly hypertrophic adenoids (> 80% obstruction of the nasopharynx), minimally enlarged palatine tonsils, normal vocal cord mobility, and significant laryngomalacia (retroflexed epiglottis with tight aryepiglottic [A-E] folds). A modified barium swallow revealed frank aspiration with thin liquids. Because of concern for obstructive sleep-disordered breathing inpatient polysomnography (PSG) was performed 8 days postextubation. It revealed near-continuous stridor and snoring. Overall moderate, but severe in rapid eye movement (REM), OSA was noted, with an overall obstructive apnea-hypopnea index (OAHI) of 7.5 events/h, REM OAHI 12.3 events/h, non-REM OAHI 5.7 events/h, associated with frequent moderate to severe desaturations (SaO_2 nadir of 74%), and severe hypercarbia (**Table 1**, **Figure 1** and **Figure 2A**). No significant central apnea was noted. In addition, the breathing pattern on PSG revealed prolonged periods of flow limitation along with choking and gasping arousals.

Figure 2—Sleep study hypnograms compared before and after adenoidectomy and supraglottoplasty.



(A) Baseline study: This hypnogram reveals severe obstructive sleep apnea (OSA) with severe desaturations and hypercarbia in rapid eye movement (REM) sleep. **(B)** After adenoidectomy and supraglottoplasty: the initial half of the hypnogram is baseline, which reveals significant OSA in REM sleep and persistent hypercarbia. **(C)** Post-Chiari decompression (after 8 months): hypnogram shows normal sleep study, no desaturation, and no apneas.

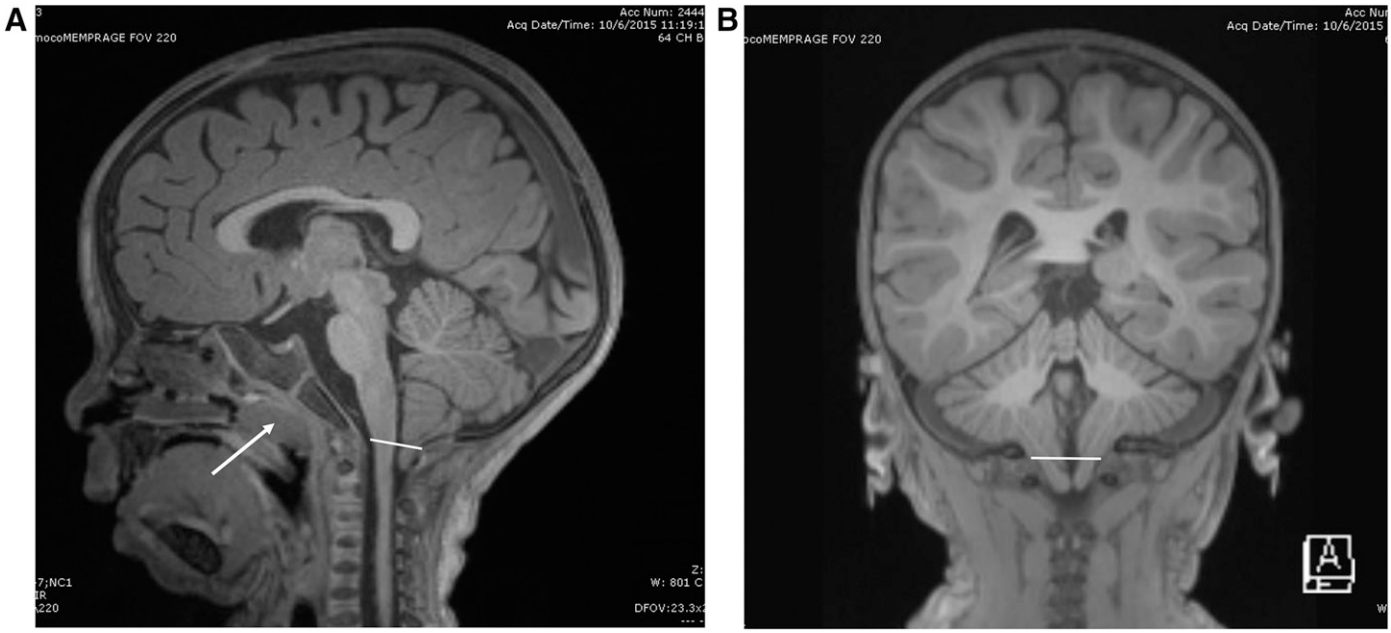
After stabilization, on day 12 of hospitalization, the patient underwent adenoidectomy and supraglottoplasty (release of tight A-E folds) due to moderate OSA with severe desaturation in conjunction with DISE, rigid airway endoscopy, and cardiac echocardiogram. During the DISE, there was mild glossoptosis, no significant lingual tonsil hypertrophy, and no pharyngomalacia. Further evaluation with DISE revealed asymmetric adduction of the vocal cords on inspiration resulting in inspiratory stridor. However, a subsequent postoperative nasopharyngoscopy done while the patient was awake 1 day later revealed return of normal vocal cord mobility. A follow-up PSG performed 9 days after surgery revealed persistent stridor, snoring, increased work of breathing, frequent flow limitations, sleep hypercarbia (CO_2 55–65 mm Hg), and REM-related severe desaturations with nadir of 80% (**Table 1**). The overall OAHI (2.2 events/h) indicated modest improvement and no central apneas were noted. However, because of persistent hypercarbia and snoring/stridor the patient underwent bilevel positive airway pressure (BPAP) titration. The patient required relatively high BPAP of 16/6 cm H20 without backup rate to control sleep-disordered breathing. On these settings, his gas exchange normalized and snoring and stridor resolved (**Figure 2B**).

The residual sleep-disordered breathing was attributed to arise from abnormal vocal cord motion (adduction on inspiration rather than abduction) during sleep. With the descent of the brainstem in ACM type 1 the vagal nuclei in the brainstem can become compressed, resulting in bilateral or unilateral vocal cord paresis and stridor. After careful consideration and review of the vocal cord mobility on the DISE video, MRI with focus on posterior fossa of the brain was ordered. The brainstem MRI revealed cerebellar tonsils that extended inferiorly to C2 (10 mm), consistent with ACM type 1, and no syrinx (**Figure 3**). A multidisciplinary team meeting concluded that this constellation of symptoms and clinical findings were consistent with symptomatic ACM. The patient underwent craniotomy with posterior fossa decompression. Postoperatively, the patient did very well, and his stridor and severity of desaturation significantly improved within 24 hours. The patient was discharged on supplemental oxygen due to sleep hypoxemia that was attributed to low pulmonary reserve due to ongoing aspiration. A chest radiograph revealed low lung volume, streaky, patchy atelectasis, prominent bronchovascular markings, and peribronchial cuffing especially on the right side (**Figure 4**). Follow-up PSG performed 1 month after decompression surgery revealed complete resolution of previously noted stridor, OSA, and sleep hypercarbia; however, mild sleep hypoxemia (SaO_2 92% to 95%) persisted with nadir desaturation of 69%. The patient's dysphagia and aspiration gradually resolved over the following 4 to 6 months and a repeat modified barium swallow study showed no aspiration and he continued to tolerate thin liquids orally. Another follow-up PSG 6 months after decompression surgery, confirmed the continued resolution of OSA (OAHI 0 events/h), no central sleep apnea, no bradypnea, normalization of sleep oxygenation (SaO_2 nadir 97% to 99% off supplemental oxygen), and normal CO_2 levels (**Figure 2C**). Oxygen therapy was then discontinued.

DISCUSSION

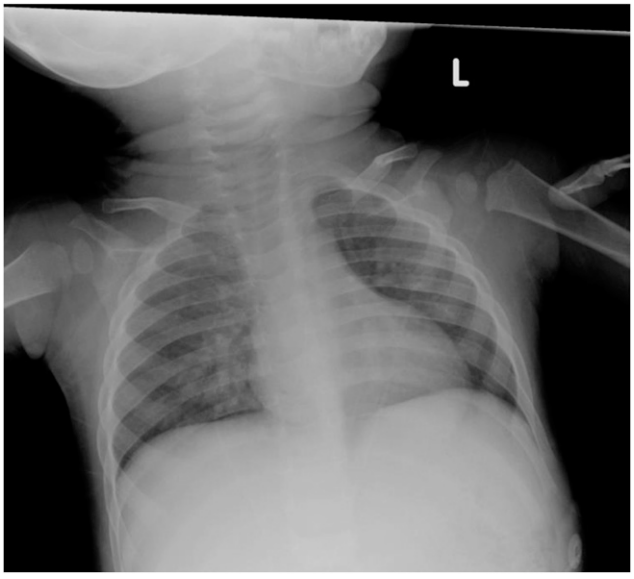
ACM was first described by Hans Chiari in 1891.[2] Since then, the evolution of modern imaging techniques and MRI has clarified the definition of ACM as an extension of the

Figure 3—Magnetic resonance imaging (MRI) of the brain.



(A) Sagittal MRI study of the brain shows the herniation of cerebellar tonsils 10 mm below the foramen magnum into the spinal canal. **(B)** Coronal MRI study showing T2 signal prolongation within the medial aspect of the right cerebellar hemisphere, consistent with a prior petechial hemorrhage.

Figure 4—Chest radiograph showing low lung volume, streaky atelectasis, bronchovascular crowding, and fluffy opacities on the right side.



cerebellar tonsils by more than 6 mm below the foramen magnum.[6,7] Given that the extent of herniation of cerebellar tonsils varies across patients, individuals with ACM may exhibit a wide range of symptoms, ranging from totally asymptomatic to headaches, dysphagia, nystagmus, and gait abnormalities.[8] ACM is grouped into three general types. Type 1 is the most common and often presents no symptoms for much of the patient's life. Type 2 often presents with meningomyelocele or other comorbid condition such as dysphagia, arm weakness, stridor, apneic spells, and aspiration.[9] Type 3 is the most severe with inferior displacement of the medulla with a high cervical or occipital encephalocele that typically contains much of the cerebellum.

ACM type 1 is usually diagnosed in the adult population because children are often asymptomatic.[10] However, many cases of pediatric ACM have been described in the literature. One study identified oropharyngeal dysphagia as the most common symptom among children with ACM type 1. Many patients with ACM present with central sleep apnea, bradypnea, and sleep hypoventilation due to pressure on the respiratory center of the medulla oblongata. Although OSA was not as common, in one study 29% of children with ACM exhibited some degree of sleep-disordered breathing.[11] Additionally, these patients with sleep-disordered breathing tend to develop neurocognitive, cardiovascular, behavioral disorders, inattention/hyperactive disorders, and academic difficulties.[12–15] Therefore, when ACM is diagnosed on MRI performed for other reasons underlying sleep disordered breathing needs to be evaluated and periodically followed up with timely treatment.

In patients with persistent OSA following adenotonsillectomy, several diagnostic evaluation techniques can be used to further assess the upper airway. One very useful tool as seen in this case is DISE. This procedure allows clinicians to precisely assess dynamic function of the upper airway and other sites of obstruction.[5] Many DISE classifications to assess the pattern of upper airway obstruction does not effectively assess vocal cord movements and hence this site can

be easily missed. In our clinical practice we routinely examine the nasopharynx, palatine tonsils, lingual tonsils, and supraglottic and glottis regions in addition to the VOTE (Velum, Oropharynx, Tongue base and Epiglottis) classification parameters during DISE. This case demonstrates the importance of assessing vocal cord mobility during DISE.[16] Vocal cord dysfunction is one key factor of ACM-derived obstructive breathing as displaced cerebellar tonsils can adversely affect the vagal nuclei in the brainstem, particularly in supine position.[17] In our case, the presence of vocal cord dysmotility was identified only during DISE but not while awake, confirming it as the primary etiology for obstructive breathing and gas-exchange abnormalities during sleep.

Management of OSA varies depending on the etiology. Although surgery is often warranted, medical management with noninvasive positive pressure ventilation or oxygen therapy is often used in patients experiencing central apneas or hypoventilation. BiPAP therapy was initiated in our patient given the degree of hypoventilation.[4] However, as the patient required relatively higher pressure for control of sleep-disordered breathing it is likely that this is due to a "fixed" pattern of obstruction because of adduction of vocal cords during sleep. However, some patients with ACM experience progressive worsening of symptoms over the course of the night, particularly during non-REM stages; if the symptoms are severe these may not be fully addressed by BiPAP. In these cases, surgical decompression can be curative.

In our patient, stridor and obstructive sleep-disordered breathing resolved after posterior fossa decompression surgery, indicating that this was directly attributed to the ACM. We know that compression of the brainstem against the foramen magnum during sleep results in altered activity of the respiratory center leading to central sleep-disordered breathing in addition to abnormality of vocal cords resulting in obstructive sleep-disordered breathing. Hence, treating ACM leads to resolution in sleep-disordered breathing and at times the improvement can be instantaneous or delayed and sometimes the mechanisms may not be very clear. However, this case illuminates both the challenge of identifying the primary site of obstruction (as vocal cord mobility was normal when awake) and the value in careful assessment of vocal cord mobility while sedated during DISE mimicking natural sleep.

CONCLUSIONS

Sleep-disordered breathing is a known but poorly evaluated comorbidity in patients with ACM. The cause of upper airway obstruction in these patients could be due to abnormal vocal cord movements that may be seen only during sleep but are normal when awake. Hence the use of DISE can help evaluate patients with ACM and persistent sleep-related breathing disorders. In such cases, a careful and thorough examination of vocal cord mobility should be performed and can help direct management of both the ACM and the breathing disturbances during sleep.

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DISCLOSURE STATEMENT

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