Pediatric Pulmonology 45:1141–1144 (2010)

Case Report

Isolated Central Sleep Apnea in Type I Chiari Malformation: Improvement After Surgery

Jessica Spence, BMR(OT),[1,2] Hans Pasterkamp, MD, FRCSC,[2,3] and Patrick J. McDonald, MD, MHSc, FRCSC[1,2]*

Summary. Sleep apnea is a rare but well-known clinical feature of disorders of the craniocervical junction. It may be obstructive or central in nature, and rarely presents without other neurological symptoms. We report the cases of two children, presenting with isolated central sleep apnea leading to a diagnosis of type I Chiari malformation. Surgical treatment resulted in a dramatic improvement in respiratory parameters during sleep, both clinically and on polysomnography. We discuss this uncommon presentation of type I Chiari malformation and suggest that it be considered in the differential diagnosis of central sleep apnea in children, as posterior fossa decompression may lead to significant clinical improvement. **Pediatr Pulmonol. 2010; 45:1141–1144.** © 2010 Wiley-Liss, Inc.

Key words: chiari malformation; sleep apnea; surgery.

Funding source: None reported.

INTRODUCTION

The type I Chiari malformation (CM-I) is a congenital brainstem abnormality characterized by caudal descent of the cerebellar tonsils below the level of the foramen magnum, resulting in crowding at the craniocervical junction. It typically presents in adulthood, most commonly with symptoms of headache, neck pain, ataxia, sensory deficit, lower cranial nerve palsies, and nystagmus. When it does present in the pediatric population, it is most frequently associated with headache, although one-third are asymptomatic.[1,2]

Given the anatomic location of the respiratory centres in the medulla, it is not surprising that compression of these structures in CM-I has been associated with sleep-disordered breathing.[3] All types of apnea—central, obstructive and mixed—have been described, with obstructive being most common.[4] Although it remains unclear how frequent sleep apnea is in CM-I, its recognition is usually associated with other neurological manifestations.[5] First described by Balk et al.[6] there are few reports of sleep apnea syndrome as the sole neurological manifestation of CM-I, both in children and adults.[5,7–9] As such, the presence of central apnea and bradypnea on polysomnography in otherwise well children merits investigation by MRI to rule out the presence of CM-I.

In this article, we describe two pediatric cases presenting with central sleep apnea as the sole symptom of CM-I. Surgical decompression was able to provide definitive treatment for these patients, eliminating the need for the use ventilatory support during sleep.

CASE 1

A 7-year-old girl was referred to our pediatric respirology clinic for investigation of sleep-disordered breathing. Her parents noticed long pauses in breathing

[1]Sections of Neurosurgery, University of Manitoba Winnipeg, Manitoba, Canada.

[2]Winnipeg Children's Hospital, Winnipeg, Manitoba, Canada.

[3]Pediatric Respirology, University of Manitoba, Winnipeg, Manitoba, Canada.

*Correspondence to: Patrick J. McDonald, MD, MHSc, FRCSC, Section of Neurosurgery, University of Manitoba, GB-126, 820 Sherbrook Street, Winnipeg, MB, Canada R3A 1R9. E-mail: pmcdonald@hsc.mb.ca

Received 30 September 2009; Revised 21 April 2010; Accepted 22 April 2010.

DOI 10.1002/ppul.21294
Published online 16 September 2010 in Wiley Online Library (wileyonlinelibrary.com).

while asleep when she was a small baby. These became more pronounced when she was 3–4 years of age. During these episodes the child would take several deep breaths and then stop breathing for 10–15 sec, resuming after either she stirred or her parents woke her up. Her previous medical history was significant for type II Diabetes Mellitus, and her body mass index (BMI) was 24.7.

Polysomnography showed a severe degree of sleep disordered breathing and an apnea–hypopnea index (AHI) of 82.2 (normally <1), with 9 hypopneas, 44 obstructive apneas, 13 mixed episodes, and 507 central apneas associated with desaturation and arousals from sleep (Table 1). A trial of continuous positive airway pressure (CPAP) was initiated and, after non-response, a neurology consult and an MRI of the brain were arranged to investigate the cause of her central apneas. In the meantime, biPAP was tried and shown to be effective in normalizing her respiratory pattern (Table 1).

MRI demonstrated a CM-I, with 2 cm of tonsillar ectopia and some signal change in the adjacent cervical spinal cord, indicative of chronic compression. She underwent occipital craniectomy, C1/C2 laminectomy, duraplasty, and coagulation of the cerebellar tonsils, which were obviously atrophic and compressing the upper cervical cord.

Immediately after surgery a clinical improvement was noted in her breathing pattern. Polysomnography, done 3 and 15 months post-operatively, confirmed this, showing improved sleep architecture (Table 1). A number of central apneas persisted, primarily during stage II sleep, but were no longer associated with desaturations below 90%. The highest end-tidal pCO_2 pre- and post-operatively was 44.8 and 46.4 mmHg, respectively. Capillary blood gas analysis at the end of sleep recordings showed a pCO_2 of 36 pre-operatively and 37 post-operatively.

The patient remains clinically improved now over 15 months after her surgery with no evidence of a sleep-related breathing disorder.

CASE 2

A 15-year-old boy was seen in the pediatric respirology clinic with concerns of sleep apnea. His mother described a 2-year history of episodes occurring repeatedly through the night, during which he would take several deep, noisy breaths and then become quiet for 15–25 sec. He had no significant previous medical history, although initial work-up showed hypercholesterolemia and a BMI of 29.0.

Polysomnography was significant for sleep disordered breathing, with an AHI of 66. The study recorded 39 hypopneas, zero obstructive sleep apneas, and 295 central apneas, most of which were associated with significant desaturation (Table 1). CPAP was started, with no response. BiPAP was initiated and he subsequently improved, reporting improved sleep and daytime alertness.

ENT examination by flexible nasolaryngoscopy revealed a normal nasal cavity and nasopharynx, small adenoid pad and no abnormality to the oropharynx or larynx itself. Nonetheless, a decision was made to proceed with tonsillectomy and adenoidectomy to obtain symptom improvement and decrease biPAP usage. Polysomnography done post-operatively showed an improvement in his AHI (from 66 to 25) and a much more stable SaO_2 with use of biPAP. In spite of this, severe central sleep apnea persisted, with associated desaturations as low as 70% (Table 1).

Further investigation included MR imaging (see Fig. 1), which showed a significant CM-I with 2.5 cm of tonsillar ectopia and indications of early syringomyelia in the cervical cord. Platybasia was also noted. Posterior fossa decompression and C1/C2 laminectomy, with dissection

TABLE 1— Polysomnography Pre- and Post-Chiari Decompression

	Patient 1				Patient 2			
	Baseline	BiPAP	3 months post-operatively	15 months post-operatively	Baseline	BiPAP	3 months post-operatively	9 months post-operatively
Weight (kg)	44.8	45	45	44	77	77	77	77
BMI	24.4	24.4	24.3	22.4	29.0	29.0	29.0	29.0
Sleep architecture								
Total sleep time (hr)	6:58	4:21	7:40	5:17	5:08	6:22	5:43	7:05
Sleep efficiency(%)	83.9%	53.5	89.9	67.4	71.9	76.5	80.7	92.8
Sleep onset latency(min)	37	81	32	24	47	19	22	17
REM latency (hr)	0:59	4:53	2:14	3:38	4:56	2:07	1:05	1:11
Respiratory variables								
Hypopneas	9	1	136	0	39	23	8	6
Apneas								
Obstructive	44	1	0	0	0	1	0	0
Mixed	13	0	0	0	7	0	0	1
Central	507	57	56	1	295	188	27	125
Apnea–hypopnea index (hr)	82.2	13.6	25.0	0.2	66.0	25.4	6.1	18.6
Mean oxygen saturation (%)	96	98	98	98	89	93	99	96
Minutes <90%	0.1	0	0	0	41.5	35.3	0	2.1

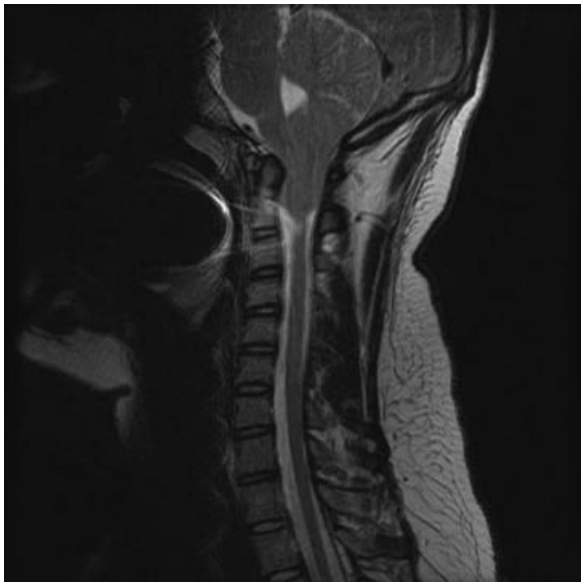

Fig. 1. Pre-operative saggital T2-weighted magnetic resonance (MR) image of the craniocervical junction illustrating 2.5 cm of tonsillar herniation below the foramen magnum.

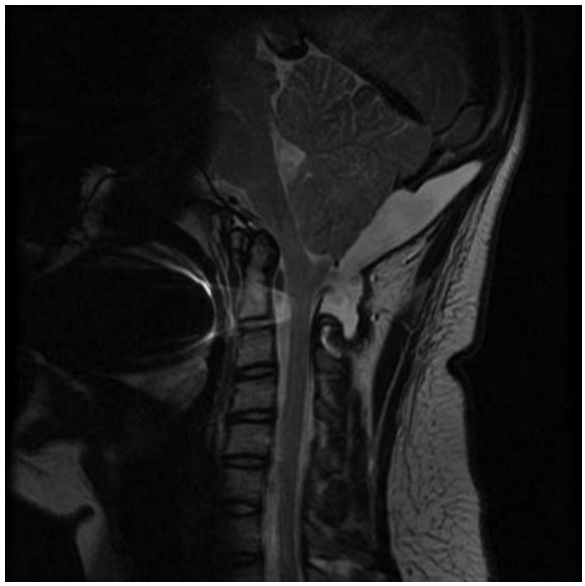

Fig. 2. Post-operative saggital T2-weighted image of the craniocervical junction in the same patient.

and coagulation of the cerebellar tonsils was done on a relatively urgent basis, as the boy's parents had begun to note periods of apnea occurring during the day. Interestingly, during surgery, the degree of compression of the cervical cord was noted to be so severe that there was actually a dorsal hump of spinal cord herniating through the two cerebellar tonsils.

After surgery, an immediate clinical improvement in the patient's breathing was noted, and polysomnography was done to confirm the safety of biPAP discontinuation. A second study, done 3 months post-operatively, confirmed this dramatic improvement, with a decrease in AHI from 25 to 6.1 and a mean SaO_2 of 99% (Table 1). End-tidal pCO_2 was 48.8 mmHg pre-operatively and 54.9 post-operatively with corresponding capillary blood gas at the end of sleep recordings revealing a pCO_2 of 47 mmHg pre-operatively and 46 post-operatively.

Now 1 year post-surgery, the patient's clinical improvement is sustained, although repeat polysomnography at 9 months showed an increase in the number of central apneas and an increase in the AHI compared to 3 months post-surgery (Table 1).

DISCUSSION

CM-I has historically been thought of as a rare disease. However, with routine use of MR imaging, it is discovered with increasing frequency. Currently, estimates of prevalence suggest rates of 0.1–0.5% with a slight female preponderance.[10]

The presence of sleep-disordered breathing in association with CM-I is well-recognized although it rarely presents without other neurological symptoms.[5] The frequency of sleep apnea/hypopnea in CM-1 is unknown but recent findings suggest it may be present in more than half of symptomatic CM-I patients. In their series, Gagnadoux et al.[11] found sleep disordered breathing in 12/16 patients presenting with symptomatic CM-I and syringomyelia, with 50% of these being central apneas. The frequent presence of sleep apnea syndrome in CM was confirmed by the work of Dauvillier et al.[4] who found that 60% of a series of 20 children (15 type I and 5 type II) had a diagnosis of sleep apnea syndrome on polysomnography.

The speculative pathophysiology underlying this manifestation is heterogenous and relates to compression of the brainstem. Resulting lesions may subsequently result in apnea through depression of respiratory afferents, medullary respiratory centres or efferents to the pharyngeal apparatus and chest wall.[4] Peripheral chemoreceptor abnormalities have been detected in some children with Type II Chiari malformation (CM-II), supporting the notion that brainstem compression may lead to afferent dysfunction.[12] Additionally, compression of nuclei associated with cranial nerves 9 and 10, responsible for control of vocal cords and pharyngeal musculature, may result in efferent dysfunction and diminished airway patency during sleep. Consistent with this, Dauvillier et al.[4] found that vocal cord paralysis was the single most important predictor of central respiratory events in CM, suggesting that all individuals with this diagnosis, especially CM-II associated with spinal dysraphism, should undergo regular laryngoscopic exam. These impairments manifest either as hypercapnia (due

to blunted bulbar chemical drive) or normo/hypocapnia (from an increased bulbar chemical drive that destabilizes the breathing pattern during sleep).

Given the relative infrequency of CM-I presenting with isolated central sleep apnea in the literature, it is difficult to make conjectures about predisposing factors. Consideration must be given to an apneic threshold that is either inherently increased or, through anatomic modification, results in decreased ventilatory responsiveness. Though typically associated with obstructive sleep apnea, the presence of both obesity and cardiovascular risk factors in both of our patients lead one to wonder whether these may play a role as well.

Nonetheless, given that brainstem compression leads to the ultimate insult, one would expect that surgical decompression would lead to an improvement in respiratory parameters of sleep. Recently, several case reports have been published supporting this approach.[4,5,7,9–11] Still, the role of surgery in children with CM-I remains difficult to assess, as most reports describe patients with significant neurological findings.[1]

It has been suggested that adults with sleep-disordered breathing due to CM-I should undergo surgery to prevent both the potentially life-threatening sequelae of sleep apnea and possible onset of other neurological symptoms.[4,8] Additionally, there is evidence that ongoing sleep disordered breathing may exacerbate the underlying neurological condition. Pasterkamp et al.[13] described an increase in intracranial pressure and development of syringomyelia secondary to obstructive sleep apnea in a patient with CM-II. Early diagnosis is particularly important, as late intervention may preclude a surgical cure.[14]

In any case, central sleep apnea and bradypnea on polysomnography in otherwise well children should raise concerns of brainstem compression and the possibility of a surgical cure. When recognized, early intervention should be considered, as ongoing sleep disordered breathing poses threats of respiratory failure, and may result in subtle developmental delay and learning difficulties because of daytime somnolence. In patients with severe sleep apnea, as our patients illustrate, decompression of the craniocervical junction can bring about significant improvement, even with long standing central sleep apnea. Although improvement post-surgery was sustained over time in the patients presented, as one of our patients has shown, central apneas can recur, highlighting the need for long-term follow-up and ongoing vigilence.

REFERENCES

1. Tubbs RS, McGirt MJ, Oakes WJ. Surgical experience in 130 pediatric patients with Chiari 1 malformations. J Neurosurg 2003; 99:291–296.
2. Novegno F, Caldarelli M, Massa A, Chieffo D, Massimi L, Pettorini B, Tamburrini G, DiRocco C. The natural history of the Chiari Type I anomaly. J Neurosurg Pediatr 2008;2:179–187.
3. Reybet-Degat O. Pathologies de la charnière cervical et apnées du sommeil. Rev Neurol 2001;157:5S156–5S160.
4. Dauvilliers Y, Stal V, Abril B, Coubes P, Bobin S, Touchon J, Escourrou P, Parker F, Bourgin P. Chiari malformation and sleep related breathing disorders. J Neurol Neurosurg Psychiatry 2007; 78:1344–1348.
5. Zolty P, Sanders MH, Pollack IF. Chiari malformation and sleep-disordered breathing: a review of diagnostic and management issues. Sleep 2000;23:637–643.
6. Balk RA, Hiller FC, Lucas EA, Scrima L, Wilson FJ, Wooten V. Sleep apnea and the Arnold-Chiarimalformation. Am Rev Respir Dis 1985;132:929–930.
7. Lam B, Ryan CF. Arnold-Chiari malformation presenting as sleep apnea syndrome. Sleep Med 2000;1:139–144.
8. Gosalakkal JA. Sleep-disordered breathing in Chiari malformation type I. Pediatr Neurol 2008;39:207–208.
9. Miralbes Terraza S, Garcia Oguiza A, Lopez Pison J, Saenz Moreno I, Jimenez Escobar V, Pena Segura JL, Marco Rived A. Central sleep apnea syndrome as the first sign of type I Chiari malformation. An Pediatr (Barc) 2008;68:277–281.
10. Speer MC, Enterline DS, Mehltretter L, Hamoock P, Joseph H, Dickerson M, Ellenbogen RG, Milhorat TH, Hauser MA, George TM. Chiari type I malformation with or without syringomyelia: prevalence and genetics. J Genet Couns 2003;12:297–311.
11. Gagnadoux F, Meslier N, Svab I, Menei P, Racineux JL. Sleep-disordered breathing in patients with Chiari malformation: improvement after surgery. Neurology 2006;66:136–138.
12. Gozal D, Arens R, Omlin KJ, Jacobs RA, Keens TG. Peripheral chemoreceptor function in children with Myelomeningocele and Arnold-Chiari malformation type 2. Chest 1995;108:425–431.
13. Pasterkamp H, Cardoso ER, Booth FA. Obstructive sleep apnea leading to increased intracranial pressure in a patient with hydrocephalus and syringomyelia. Chest 1989;95:1064–1067.
14. Tsara V, Serasli E, Kimiskidis V, Papagianopoulos S, Katsaridis V, Fylaktakis M, Christaki P, Kazis A. Acute respiratory failure and sleep-disordered breathing in Arnold-Chiari malformation. Clin Neurol Neurosurg 2005;107:521–524.