Novel treatment (new drug/intervention; established drug/procedure in new situation)

Chiari malformation and central sleep apnoea: successful therapy with adaptive pressure support servo-ventilation following surgical treatment

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Summary

Sleep apnoea is a common disorder with significant morbidity. It is categorised into obstructive and central sleep apnoea. There are a variety of conditions associated with central sleep apnoea ranging from cardiac failure to structural brain anomalies. We herein report a case of 57-year-old woman with Chiari malformation associated with significant sleep-disordered breathing. There was a family history of Chiari malformation. Although neurosurgical intervention had a significant impact on apnoea hypopnoea index (AHI) with a reduction from 81/h preoperatively to 22.1/h after the surgical treatment, it failed to cure the sleep disorder breathing completely and adaptive non-invasive servo-ventilation had a dramatic effect on symptoms and normalised the AHI. Although, central sleep apnoea is associated with a number of common disorders, this case illustrates that there may be an unusual cause. Moreover, adaptive pressure support servo-ventilation may provide a promising treatment option following surgical correction of Chiari malformation.

BACKGROUND

Sleep-disordered breathing is a significant global health issue. Recent epidemic of obesity has played a significant role in the rise of sleep apnoea. There are two broad categories of sleep apnoea, obstructive and central sleep apnoea. Moreover, a mixture of these categories is frequently encountered. Although obstructive sleep apnoea is a more common variety, central sleep apnoea may be encountered in the context of cardiac failure, acromegaly^{1 2} and renal failure.³ Herein, we report a case of central sleep apnoea, the aetiology of which is described very rarely. Furthermore, a novel ventilatory strategy following surgical correction has been utilised, which to our knowledge, has not been reported in Chiari malformation before.

CASE PRESENTATION

A 57-year-old woman was referred to our sleep clinic with a history of snoring and witnessed episodes of apnoea. She also complained of tiredness, lethargy and lack of concentration and motivation. She described waking up coughing as though she was choking. Her snoring was severe; she was restless in bed and woke several times during the night to go to the toilet. Her sleep hygiene was that of a normal pattern and had no problems falling asleep. She denied any orthopnoea or paroxysmal nocturnal dyspnoea. Epworth score was elevated at 18 of 24.

She was a lifelong non-smoker and consumed minimal amount of alcohol. Basic ear, nose and tongue examination showed a large palate and a crowded pharynx. Her weight was 99.6 kg with a body mass index of 35.3. Collar size was measured at 44 cm. There was no evidence of cardiac failure and the rest of the systemic examination was unremarkable.

INVESTIGATIONS AND MANAGEMENT

Overnight pulse oximetry showed evidence of significant 4% desaturation rate of 38/h with minimum and mean oxygen saturation of 75% and 93%, respectively. This oximetry results in the context of her symptoms supported a diagnosis of severe obstructive sleep apnoea. Hence she was started on a trial of continuous positive airway pressure (CPAP) therapy (Remstar Auto M series CPAP machine). Her compliance was good with an average usage of 6 h per night every night. However, apnoea hypopnoea index (AHI) remained elevated at 36.7 with a baseline CPAP pressure of 7 cm H₂O. Epworth score remained elevated at 14/24. In view of the high CPAP pressure required she was started on bilevel positive airway pressure (BIPAP) with a Remstar BIPAP Auto M series machine on auto mode to allow for a higher pressure to be delivered. Inspiratory positive airway pressure (IPAP) and expiratory positive airway pressure were 25 and 10 cm H₂O, respectively. Despite good compliance with an average usage of 8 h every night, AHI increased to 52 and symptoms remained persistent. In view of poor response to CPAP and BIPAP, a possibility of central or complex sleep apnoea was considered and a sleep study was performed. The sleep study showed AHI of 81/h with predominantly central apnoeas (99%). An echocardiogram to evaluate for cardiac failure showed mild pulmonary hypertension in keeping with nocturnal hypoxaemia. However, there was no evidence of ventricular dysfunction.

In view of poor response to CPAP and BIPAP, the patient was started on a trial of adaptive servo-ventilation (ASV). It can treat a mixture of periodic and complex breathing by monitoring and reacting to changes in peak expiratory flow thus preventing cyclical cheyne stoke respiration. Patient reported to be feeling more comfortable on the auto SV as opposed to CPAP therapy. A further

sleep study while using ASV showed an improvement of AHI to 10.5/h with persistent hypopnoeas.

At next outpatient review, the patient informed that her son had been diagnosed with Arnold-Chiari malformation. In view of an association of Arnold-Chiari malformation with sleep-disordered breathing (specifically central sleep apnoea), an MRI scan of brain (figure 1) was arranged. MRI scan demonstrated significant Chiari type 1 malformation and a syrinx in the spinal cord.

She was referred for neurosurgical intervention and underwent craniocervical decompression for the correction of brain malformation. A follow-up sleep study a couple of weeks after surgical correction, while using ASV, showed complete resolution of sleep apnoea. The patient reported a dramatic improvement in daytime sleepiness and concentration and she was waking up less frequently during the night. To evaluate if the subjective and objective improvement of central sleep apnoea was related to surgical treatment alone, another sleep study was undertaken without ASV. It demonstrated that AHI had increased to 22. Serial sleep studies (table 1) from the outset showed that central sleep apnoea in this patient improved but not cured by surgery alone and ASV had a substantial benefit following surgical correction.

DISCUSSION

Chiari malformations consist of a downward displacement of the cerebellar tonsils through the foramen magnum and were first described by John Cleland in 1883⁴ and subsequently were classified in 1891 into four

Figure 1 MRI of brain showing downward displacement of cerebellar tonsil (arrow) at the level of foramen magnum consistent with type 1 Chiari malformation.

Table 1 Sleep studies during the course of investigation and management of the patient

Measure	Baseline	Auto SV	Auto SV postoperation	No NIV postoperation
AI	68	0.1	0.1	8.4
AHI	81	10.5	0.7	22.1
Total apnoeas	523	1	1	81
Central	522 (99%)	1 (100%)	1 (100%)	69 (85%)
Obstructive	1 (0%)	0	0	3 (4%)
Mixed	0	0	0	9 (11%)
Hypopnoeas	103	105	7	132
Duration mean (max)	23 (46)	16 (48)	14 (18)	17 (59)
Mean SpO ₂	91%	95%	94%	93%
4% Dip rate	79	35	9.6	39.5

AHI, apnoea hypopnea index; AI, apnoea index; NIV, non-invasive ventilation; SV, servo-ventilation.

groups.⁵ Type 1 Chiari malformation is the most common variety with caudal prolapse of cerebellar tonsils to the level of upper cervical spinal canal. Type 2 malformation is a more severe form with displacement of cerebellar vermis and tonsils and a spinal myelomeningocele.⁶ Type 3 is rare and is associated with a high cervical or occipital encephalocele along with inferior displacement of brainstem into the spinal canal. Type 4 refers to cerebellar hypoplasia without significant relation to other Chiari malformations. There have been a handful of cases of central sleep apnoea in relation to Chiari malformation.^{7–9} The exact mechanism of sleep disordered breathing secondary to this malformation is not known to a great extent. However, it may either be related to upper airway dysfunction resulting in obstructive sleep apnoea or disruption of central respiratory drive manifesting as central sleep apnoea. Furthermore, mechanical effects of cerebellar herniation on respiratory centre in medulla may be another mechanism of sleep-related apnoeic spells in this disorder.

This case demonstrates the effectiveness of servo-assisted ventilation in a patient with severe central sleep apnoea in association with brain malformation. The patient described in this case report had minimal subjective and objective benefit by CPAP or BIPAP mode of non-invasive ventilation prior to surgical treatment of Chiari malformation. In view of the failed trial of these ventilatory modes, ASV trial was attempted. ASV is a mode of ventilation with automated sensing of normal breaths and assures to avoid artificial ventilation during the periods of normal breaths. Hence, it results in minimising the likelihood of cyclical or ataxic breathing and is considered a novel mode of ventilation in selected cases of sleep apnoea. There is evidence of effectiveness of ASV in opioids-induced central sleep apnoea¹⁰; however, its effectiveness in complex sleep apnoea and mixed apnoeas is not known to a great extent.

In conclusion, this case highlights the importance of considering structural brain disorders as a cause of sleep-disordered breathing particularly if there is a family history of brain malformation and a poor response to initial CPAP therapy. Moreover, it highlights the effectiveness of ASV in treating central sleep apnoea related to Chiari malformation following surgical correction, which to our knowledge has never been reported in the literature before.

Learning points

- Chiari malformation may present as central sleep apnoea particularly if there is a family history of this structural brain disorder.
- Sleep-disordered breathing associated with Chiari malformation may be resistant to conventional mode of ventilation that is, continuous positive airway pressure and bilevel positive airway pressure.
- Surgical correction of the brain anomaly along with adaptive pressure support servo-ventilation offers a promising treatment option with immediate improvement in daytime sleepiness and apnoea hypopnoea index.

Competing interests None.

Patient consent Obtained.

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