The Journal of Laryngology and Otology
April 1995, Vol. **109**, pp. 328–330

Treatment of Robin sequence with nasal CPAP

PAUL C. DEEGAN, M.B., BRIAN MCGLONE, M.B., WALTER T. MCNICHOLAS, M.D.

Abstract
A 12-year-old schoolgirl presented with severe obstructive sleep apnoea due to the Robin sequence. The sleep apnoea, together with the associated findings of daytime sleepiness, nocturia, right heart strain and growth retardation, were successfully reversed by nasal CPAP therapy. This therapy allows postponement of a decision concerning corrective surgery until after full growth has occurred.

Key words: Pierre Robin syndrome; Sleep apnoea syndromes; Positive pressure respiration

Introduction

Obstructive sleep apnoea (OSA) is associated with recurrent episodes of upper airway (UA) obstruction, hypoxia and arousal from sleep. It can cause excessive daytime sleepiness and secondary cardiopulmonary complications, including cor pulmonale (Guilleminault *et al.*, 1981; Potsic, 1989). In childhood, OSA is most commonly associated with adenoid and tonsillar enlargement (Guilleminault *et al.*, 1981; Potsic, 1989), but may also be due to the Robin sequence of micrognathia, posterior displacement of the tongue and cleft palate (Sadewitz, 1992). Most of the literature on the management of Robin sequence focuses on the first year of life, especially the neonatal period, with little attention paid to the older child and adolescent (Sadewitz, 1992; Sher, 1992). We describe an adolescent patient, with Robin sequence complicated by severe OSA, who was successfully managed with nocturnal nasal continuous positive airway pressure (NCPAP).

Case history

A 12 years and three-month-old schoolgirl with Robin sequence was referred to this department for assessment of sleep-disordered breathing. She had had a history of habitual snoring since infancy, complicated by the development of observed pauses in respiration during sleep and associated threshing body movements over the 12 months prior to presentation. A cleft palate repair had been performed at 11 months of age, a tonsillectomy/adenoidectomy at five years, and palatoplasty two years later to correct a speech disturbance.

The patient complained of chronic fatigue and daytime sleepiness, frequently falling asleep while watching television, reading or in the classroom. Despite this she managed to be top of her class in school. She also reported nocturia up to four times/night, occasional nocturnal enuresis and a night-time cough. Her weight was 29 kg (c. 3rd centile) and height 150 cm (c. 50th centile). Apart from prominent micrognathia (confirmed on lateral cephalometrograph) and a resting tachycardia, physical examination was normal. Her ECG confirmed sinus tachycardia (110-115/minute) and also showed p. pulmonale, both suggesting right heart strain.

Initial overnight ear oximetry (Ohmeda Biox 3700e; Ohmeda, Louisville, Co., USA) demonstrated recurrent profound O_2 desaturations throughout the night (Figure 1). She then had a full sleep study in the sleep laboratory using standard methodology described elsewhere (Spier *et al.*, 1986). The results of this study (baseline) are summarized in Table I. She demonstrated severe OSA with 49 obstructive apnoeas (no movement of air through the upper airway (UA), despite chest and abdominal inspiratory effort) and hypopnoeas (tidal volume < 50 per cent baseline, with associated O_2 desaturation $\geq$4 per cent) per hour of sleep. She had very poor quality sleep with no slow wave sleep (SWS) or rapid eye movement (REM) sleep.

She was commenced on NCPAP (REMstar ™; Respironics Inc., Murrayville, PA, USA) with the pressure level gradually titrated upwards over successive nights until normal overnight oximetry was achieved at a pressure level of 14 cm H_2O. This was associated with an immediate improvement in energy and daytime sleepiness. A follow-up sleep study on NCPAP was carried out after six months of continuous therapy at home (Figure 1 and Table I). All apnoeas and hypopnoeas were abolished, oxygen saturation was within normal limits and she had adequate amounts of SWS and REM sleep.

After 18 months home therapy on NCPAP, she remained well with a significant growth response, her height now being 157.2 cm and weight 43 kg (c. 25th centile). She had no further daytime sleepiness, nocturia or nocturnal enuresis. Her ECG had reverted to normal. A further overnight study demonstrated that the NCPAP pressure could be reduced to 10 cm H_2O, without reappearance of apnoea.

Discussion

In Robin sequence, upper airway (UA) obstruction is due to posterior displacement of the tongue into the oropharynx, resulting in impaired action of the genioglossus, an important UA dilating muscle (Sher, 1992). This

From the Department of Respiratory Medicine and the Respiratory Sleep Laboratory, University College and St Vincent's Hospital, Dublin, Ireland.
Accepted for publication: 18 November 1994.

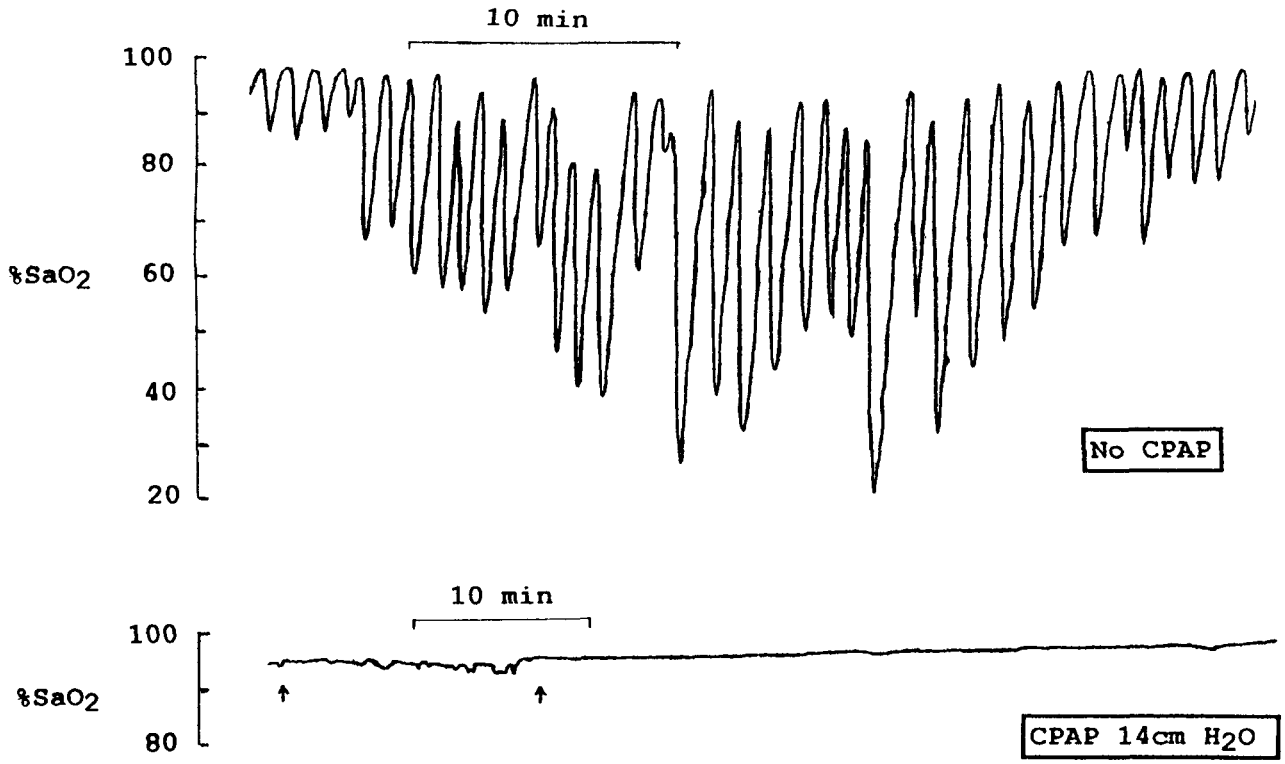

FIG. 1.

Sample of overnight oximetry tracing on screening at night (upper panel). Sample from follow-up night with nasal CPAP (lower panel). Section between the arrows represents a period of REM sleep.

has major consequences in the supine posture during sleep, when there is reduced activity of UA dilating muscles.

Treatment of the neonate is directed towards preventing feeding difficulties, pneumonia and asphyxia, and severe cases may require tracheostomy and gastrostomy (Sadewitz, 1992; Sher, 1992). The condition tends to improve after the neonatal period, particularly in those infants with isolated (i.e. nonsyndromic) micrognathia, who appear to exhibit mandibular 'catch-up growth' (Spier *et al.*, 1986). Older children and adolescents tend to have only mild sleep disordered breathing (Spier *et al.*, 1986). However, it is recognized that palatoplasty, carried out to correct speech difficulties, can lead to serious UA obstruction (Jackson *et al.*, 1976; Potsic, 1989).

In the past OSA of the severity reported here would have required tracheostomy or mandibular advancement (Guilleminault *et al.*, 1981). However, in a girl of this age, full bony and dental growth would not have occurred, and thus mandibular surgery at this stage would be suboptimal.

Nasal CPAP (Sullivan *et al.*, 1981) is recommended for moderate to severe OSA in adults, but is not widely used in children with OSA. Home use was readily accepted by our patient, with marked clinical improvement and we anticipate full physical and intellectual development. Nasal CPAP is effective only if used continuously, and she may therefore require lifelong treatment. However, the condition may ameliorate as she grows further, or alternatively corrective surgery may be considered when she reaches full bone and dental growth.

Parents may consider snoring and breath-holding normal for their child and thus not volunteer this information. A careful history and physical examination, with questions directed to elicit symptoms of night-time UA obstruction should be performed where there is a

TABLE I
SUMMARY OF SLEEP STUDIES

	Baseline	NCPAP 14 cm H_2O
Time in bed (TIB) (minutes)	432	414
Time asleep (TST) (minutes)	236	377
Stage (minutes) Awake	196	37
I	32	36
II	204	216
III	0	19
IV	0	68
REM	0	38
Sleep efficiency (TST/TIB) percentage	55	91
Apnoea/hypopnoea frequency*	49	0
Arousals*	19	1
Desaturation frequency*	47	0
Lowest O_2 saturation percentage	30	92

*Per hour of sleep; REM = rapid eye movement; Arousals = arousal pattern on EEG.

https://doi.org/10.1017/S0022215100130051 Published online by Cambridge University Press

history of UA and/or mandibular problems. Simple overnight oximetry can be a useful screening tool.

There are very few reports of the long-term use of NCPAP in children and adolescents (Ryan *et al.*, 1990), and none detailing such effectiveness in reversing the underlying clinical manifestations of sleep apnoea in this age group over a period as long as 18 months. The case reported here clearly demonstrates how a commercially available NCPAP device provides a simple noninvasive method of treatment that facilitates full physical and intellectual development. It will therefore be possible to postpone a final decision about corrective treatment until the optimum time.

References

Guilleminault, C., Korobkin, R., Winkle, R. (1981) A review of 50 children with obstructive sleep apnea syndrome. *Lung* **159:** 275–287.

Jackson, P., Whitaker, L. A., Randall, P. (1976) Airway hazards associated with pharyngeal flaps in patients who have the Pierre Robin syndrome. *Plastic and Reconstructive Surgery* **58:** 184–186.

Potsic, W. P. (1989) Sleep apnea in children. *Otolaryngologic Clinics of North America* **22:** 537-544.

Ryan, C. F., Lowe, A. A., Fleetham, J. A. (1990) Nasal continuous positive airway pressure (CPAP) therapy for obstructive sleep apnea in Hallermann-Streiff syndrome. *Clinical Pediatrics* **29:** 122–124.

Sadewitz, V. L. (1992) Robin sequence: changes in thinking leading to changes in patient care. *Cleft Palate-Craniofacial Journal* **29:** 246–253.

Sher, A. E. (1992) Mechanisms of airway obstruction in Robin sequence: implications for treatment. *Cleft Palate-Craniofacial Journal* **29:** 224-231.

Spier, S., Rivlin, J., Rowe, R. D., Egan, T. (1986) Sleep in Pierre Robin syndrome. *Chest* **90:** 711–715.

Sullivan, C. E., Issa, F., Berthon-Jones, M., Eves, L. (1981) Reversal of obstructive sleep apnoea by continuous positive airway pressure applied through the nares. *Lancet* **i:** 862–865.

Address for correspondences:
Dr Walter McNicholas,
Department of Respiratory Medicine,
St Vincent's Hospital,
Elm Park,
Dublin 4,
Ireland.

Fax: 353–1–2697949

https://doi.org/10.1017/S0022215100130051 Published online by Cambridge University Press