

Domiciliary ventilation in neuromuscular disorders - when and how?

Introduction

Respiratory failure used to be regarded as a pre-terminal complication of neuromuscular disorders and respiratory support was usually withheld for both practical and ethical reasons. Modern developments, particularly new techniques of non invasive ventilation, have radically altered this view.

Respiratory failure develops in neurological disorders when the load on the respiratory pump exceeds its capacity. The arterial PCO₂ rises as the alveolar ventilation falls and the PO₂ drops in proportion. The respiratory pump fails during sleep before wakefulness¹, and this usually occurs when the vital capacity falls to around 1 litre. In non rapid eye movement (NREM) sleep the fall in respiratory drive and the increase in upper airway resistance both tend to reduce alveolar ventilation. In rapid eye movement (REM) sleep the loss of tone in all the respiratory muscles, except the diaphragm, predisposes to apnoeas, especially if the diaphragm itself is weak.

VENTILATORY TECHNIQUES

The management of neuromuscular respiratory problems has been revolutionised by the development of non invasive ventilatory support techniques, although these were initially developed for acute poliomyelitis as long ago as the 1920s. Ventilatory support is usually only required during sleep, but almost continuous support is required if the respiratory muscle weakness is severe as in high cervical spinal cord injuries, in the later stages of motor neurone disease, or Duchenne's muscular dystrophy. It is essential to fully educate the patient and the carers and to provide a home care package which is sustainable bearing in mind the current and projected disabilities of the patient.

1. Mask Ventilation

This is usually the treatment of choice for domiciliary ventilatory support. Both pressure and volume preset ventilators are usually effective and positive end expiratory pressure (PEEP) is only required if there are also obstructive sleep apnoeas. Supplemental oxygen should not be required unless the arterial PO₂ cannot be normalised by ventilatory support alone². A nasal mask



Figure 1. Nasal Mask Ventilation

(fig 1) is usually preferable to a full face mask or a mouth-piece. Complications such as nasal ulcers, mask displacement, air leaks or upper airway obstruction may arise.

2. Negative Pressure Ventilation

A cuirass or jacket (poncho) type of negative pressure ventilator (fig 2) is preferable to the much larger tank (iron lung) ventilators for long-term use³. Both these techniques however have the disadvantage that the subjects have to lie on their back throughout the night and have a restricted range of movements⁴. There may be difficulties in fitting a cuirass or jacket to patients with a severe scoliosis secondary to muscle weakness. Negative pressure ventilation is usually used in the controlled rather than triggered mode, so that incoordination between the patient and ventilator may develop and lead to upper airway obstruction.

c. Tracheostomy Ventilation

This is required if there is upper airway obstruction, if the airway needs to be protected because of bulbar weakness, or if ventilatory support is required virtually continuously. A cuffed tube protects the airway, but the voice is lost unless the cuff can be partially or totally deflated. Tracheostomy ventilation, while effective, impairs the quality of life more than non invasive ventilation and requires a higher level of care in the home. Nevertheless it can be used successfully, particularly in those with high cervical spinal cord injuries and occasionally in motor neurone disease, although in this disorder mask ventilation is preferable whenever it can be applied successfully⁵.

d. Phrenic Nerve Stimulation (Diaphragmatic Pacing)

Phrenic nerve pacing is only indicated if the phrenic nerve is intact, diaphragm muscle function is not permanently impaired and respiratory mechanics are not grossly abnormal⁶. It is most valuable with high cervical spinal cord lesions where it can significantly increase the quality of life, mobility and speech compared with conventional tracheostomy ventilation. It can also be effective when brain stem disorders cause central alveolar hypoventilation, but simpler alternatives such as mask ventilation are usually preferable. The pacemaker requires surgical implantation, preferably in the thorax rather than the neck, and there is a small risk of phrenic nerve damage either at implantation or during pacing. Phrenic nerve pacing may induce vocal cord adduction and a tracheostomy usually needs to be retained.

INDICATIONS FOR VENTILATION

The decision about when and how to initiate domiciliary ventilation requires close collaboration with a respiratory specialist experienced in ventilatory support. It is important that access to an appropriate level of care is also available during acute intercurrent illnesses, such as chest infections, and to handle any problems that may arise with domiciliary ventilation.

a. Elective

The main indications for elective ventilatory support are shown in Table 1. 'Prophylactic' ventilatory support for



Dr John Shneerson is Consultant Physician and Director of the Respiratory Support and Sleep Centre at Papworth Hospital which is one of the largest units dealing with respiratory complications of neuromuscular disorders in the UK. His specialist interests are the physiology and consequences of respiratory failure in neuromuscular disorders and the development and assessment of new forms of treatment.

“ Non invasive ventilation not only normalises the blood gases during the night., but also during the day. Breathlessness, ankle swelling, daytime sleepiness and exercise ability all improve. ”



Figure 2. Cuirass Ventilation

asymptomatic patients who have not yet developed respiratory failure has been shown not to increase survival in Duchenne's muscular dystrophy, possibly because of complacency by the patient and family once ventilatory support is initiated so that help was not sought during acute infective exacerbations⁷.

Occasionally patients require ventilatory support to relieve orthopnoea at night in the absence of any abnormality in the blood gases either during the day or at night due to bilateral diaphragmatic paralysis. Bilateral diaphragmatic paralysis is a feature particularly of motor neurone disease, extensive multiple sclerosis, poliomyelitis, muscular dystrophies and some myopathies such as acid maltase deficiency. It can be diagnosed by the presence of orthopnoea, paradoxical inward inspiratory abdominal movement in the supine position and a fall in vital capacity on changing from a sitting to the supine position of around 50%.

TABLE 1

Indications for Elective Ventilatory Support

- Raised arterial PCO₂ during the day usually with symptoms such as sudden awakenings from sleep, irregular respiratory pattern during sleep, early morning bifrontal headaches, excessive daytime sleepiness or signs of right heart failure
- Nocturnal hypoventilation with normal waking blood gases
- Sleep disruption due to inability to lie flat even with normal night-time and waking blood gases – usually due to bilateral diaphragmatic paralysis

b. Acute

Patients with neuromuscular disorders often develop acute respiratory failure during, for instance chest infections. Ethical dilemmas may arise as to whether or not to proceed to emergency intubation and ventilation. The previous quality of life, the potential for long-term ventilatory support to improve this and the ability of the patient and the carers to cope with domiciliary ventilation all need to be taken into consideration. It is often possible to wean the subject onto a non invasive technique and to close the tracheostomy, although this may need to be retained in the long term.

OUTCOMES

Non invasive ventilation not only normalises the blood gases during the night while it is being used, but also during the day⁸. Improvement in blood gases is usually seen within a few days and can be maintained for many years, unless the neuromuscular disorder is progressive. Breathlessness, ankle swelling, daytime sleepiness and exercise ability all improve. The survival in non progressive disorders is over 80% at 5 years after starting treatment⁹.

In progressive neuromuscular disorders the outcome is determined as much by the deterioration of the underlying condition, especially bulbar function as by the adequacy of ventilatory support. Nevertheless in conditions such as motor neurone disease and Duchenne's muscular dystrophy there may be considerable symptomatic benefit and improvement in the blood gases until the condition reaches its later stages. Ventilatory support should not be withheld solely because of fear of progressing from non invasive to tracheostomy ventilation or fear of becoming virtually ventilator dependent since there are various strategies to avoid these situations if they are felt to be inappropriate.

References:

1. Bye TP, Ellis ER, Issa FG, Donnelly PM, Sullivan CE. *Respiratory failure and sleep in neuromuscular disease.* Thorax 1990;45:241-7
2. Bach JR. *Update and perspectives on noninvasive respiratory muscle aids part I: the inspiratory aids.* Chest 1994;105:1230-40
3. Shneerson J. *Non-invasive domiciliary ventilation: negative pressure techniques.* Thorax 1991;46:131-5
4. White JES, Drinnan MJ, Smithson AJ, Griffiths CJ, Gibson GJ. *Respiratory muscle activity and oxygenation during sleep in patients with muscle weakness.* Eur Respir J 1995;8:807-14
5. Oppenheimer EA. *Amyotrophic lateral sclerosis: care, survival and quality of life on home mechanical ventilation.* In: Robert D, Make BJ, Leger P, Goldberg AI, Paulus J, Willig T-N, eds. Home mechanical ventilation. Paris: Arnette-Blackwell, 1995,249-60. (proceedings of the 4th international conference on home mechanical ventilation, Lyon, 1993).
6. Moxham J, Shneerson JM. *Diaphragmatic pacing.* Am Rev Respir Dis 1993;148:533-6
7. Raphael JC, Chevre S, Chastang C, Bouvet F. *Randomised trial of preventive nasal ventilation in Duchenne muscular dystrophy.* Lancet 1994;343:1600-4
8. Jimenez JFM, Escuin JS de Cos, Vicente CD, Valle MH, Otero FF. *Nasal intermittent positive pressure ventilation.* Chest 1995;107:382-8
9. Simonds AK, Elliott MW. *Outcome of domiciliary nasal intermittent positive pressure ventilation in restrictive and obstructive disorders.* Thorax 1995;50:604-9

Correspondence to:
Dr John Shneerson, Consultant
Physician, Papworth Hospital,
Papworth Everard, Cambridge
CB3 8RE