Is chronic respiratory failure in neuromuscular diseases worth treating?

Respiratory failure develops in neurological disorders when the load on the respiratory pump exceeds its capacity. Alveolar ventilation falls, the pCO₂ rises, and the pO₂ falls proportionately. The respiratory pump may function satisfactorily initially during wakefulness and at this stage it may be only during sleep that it fails. During non-rapid eye movement sleep the reduction in respiratory drive and increase in upper airway resistance lead to a decrease in alveolar ventilation with a reduced baseline oxygen saturation and higher pCO₂. In rapid eye movement sleep not only are these changes magnified but the loss of tone in respiratory muscles other than the diaphragm leads to further underventilation and reliance on the diaphragm. Prolonged apnoeas and oxygen desaturations develop if this is weak.²³ Initially the respiratory drive is maintained,³ except in disorders of the brain stem and high cervical cord, and failure of the respiratory pump is due to a reduction in respiratory muscle strength or endurance. This is usually global and becomes significant when the vital capacity falls to around 1 litre or 25% predicted.³

Occasionally muscle weakness is selective. The most important example is bilateral diaphragmatic paralysis, which is a feature of motor neuron disease, extensive multiple sclerosis, poliomyelitis, muscular dystrophies, and some myopathies such as acid maltase deficiency. It may be manifested by orthopnoea, paradoxical inward abdominal movement during inspiration, and a fall in vital capacity on changing from the sitting to the supine position.⁴ Apart from the vital capacity the most useful screening tests for respiratory muscle weakness are the maximal inspiratory and expiratory mouth pressures. If involvement of the diaphragm is suspected more invasive investigations—including transdiaphragmatic pressures measured during voluntary deep inspirations, or sniffs, or after percutaneous electrical stimulation of the phrenic nerves or magnetic stimulation of the cervical nerve roots—may be of value.⁷

Respiratory function may also be severely compromised by abnormalities of the upper airway. These may be obvious, such as vocal cord abductor paralysis, but are often more subtle. Weakness of the dilator muscles of the upper airway predisposes to obstructive sleep apnoeas, which may present with snoring-like noises at night and increasing daytime somnolence. Upper airway obstruction or narrowing is especially important when the chest wall muscles are also weak and are unable to compensate for the increased work of breathing through the narrow airway.⁸⁹ Impairment of the swallowing mechanism or of coughing may result in difficulty in clearing tracheobronchial secretions or in aspiration into the lungs. These problems not only put the patient at risk of pneumonia but also increase the work of breathing by raising the airflow resistance and reducing the lung compliance.

The symptoms of these neurorespiratory disorders differ from the classic symptoms of lung disease such as cough, haemoptysis, and wheeze. The nocturnal respiratory disturbances cause sudden arousals from sleep, snoring-like noises, excessive daytime sleepiness, often with a deterioration in memory, changes in personality, mental and physical fatigue, and early morning headaches. Worsening breathlessness on exertion and ankle swelling are also common features.

The management of neuromuscular respiratory problems has been revolutionised by the development of long term ventilatory support techniques, especially non-invasive ventilation. These were initially developed for acute poliomyelitis. The first effective design was the tank ventilator or iron lung, which was introduced by Drinker and McKhann in 1928.¹⁰ Since then negative pressure ventilators have been simplified and the tank ventilator now only has a role in acute exacerbations and in weaning after intubation. The smaller cuirass and jacket designs, which enclose only the chest and abdomen, both have a rigid airtight framework from within which the air is evacuated by a negative pressure ventilator or pump.¹¹ Neither requires any apparatus around the patients' airways but patients have to lie on their backs while being ventilated. The cuirass has to be constructed individually to fit each patient, especially if a scoliosis is present. Negative pressure systems may induce obstruction of the upper airway particularly if the upper airway dilators are weak and unable to counteract the negative pressure generated by the ventilator.¹² A drawback with negative pressure ventilation is that the pumps are not as complex as modern positive pressure ventilators. They are all pressure preset and have fixed inspiratory and expiratory times. Their pressure wave form and the ability to alter their flow rate vary considerably and determine the degree to which they are able to compensate for leaks and to which they predispose to upper airway obstruction.¹³

The stimulus for the development of long term positive pressure ventilation was the 1952 Copenhagen poliomyelitis epidemic in which improved survival was obtained with a cuffed tracheostomy tube which protected the airway.¹⁴ Bulbar dysfunction and upper airway obstruction are still indications for a tracheostomy but in
the home this requires an extra level of care above what is needed with non-invasive techniques. Over the past 10 years the indications for tracheostomy ventilation have reduced as the design of nasal, mouth, and face masks and similar appliances has advanced.11-16 These do not protect the airway and can lead to damage to pressure areas on the face17 but they can be used effectively to ventilate most patients with chronic neurological respiratory failure and have the advantage that they can be readily applied and removed. This makes them, like the cuirass and jacket, ideal for nocturnal use in the home to support respiration during sleep, when it is most vulnerable. As with negative pressure ventilators there can be problems with coordination, upper airway obstruction, and leaks,18 but careful attention to the selection of the ventilator and the ventilator settings usually enables these difficulties to be overcome.

These non-invasive techniques suit most neurological patients with chronic respiratory failure but a minority require other forms of treatment. Phrenic nerve pacing is the best known but is only indicated if the phrenic nerve is intact, diaphragm muscle function is not permanently impaired, and the respiratory mechanics are not grossly abnormal.19 Its main value is in high cerebral tetraplegia, where it can significantly increase the quality of life, mobility, and speech compared with conventional tracheostomy ventilation. It is also effective in brain stem disorders causing central alveolar hypoventilation but simpler alternatives are usually available for these patients. The pacemaker requires surgical implantation, usually in the thorax, but occasionally in the neck. There is a small but definite risk of phrenic nerve damage at implantation or during pacing but diaphragm fatigue should be uncommon with modern pacing methods.

The benefits of ventilatory support are due to the combined effects of several interrelated mechanisms. The respiratory drive increases, probably both through an improvement in sleep quality20 and a reduction in CSF bicarbonate which resets the ventilatory response to CO2. Respiratory muscle strength and endurance may improve and the work of breathing may fall as a result of an increase in the chest wall and lung compliance. Within a few days the arterial blood gases improve both during the day and at night, even with treatment only at night, and the new levels are maintained in non-progressive neurological disorders for several years.21-25 Breathlessness, ankle swelling, daytime sleepiness, and exercise ability26 may all improve. The patients are often able to return to work or continue schooling. There have been no large controlled studies of survival in these disorders with and without treatment but with ventilatory support the five-year survival in slowly and non-progressive disorders such as poliomyelitis, muscular dystrophies, and myopathies is over 80%.27,28

If non-invasive ventilation is so effective should it be used before respiratory failure has developed to delay its onset and reduce the risk of complications? This question has been considered for Duchenne’s muscular dystrophy in a recent large French trial.29 Patients were randomised either to nasal ventilation or conventional treatment but the survival was worse in the ventilated group. This may have been because nasal ventilation gave the patients and family a false sense of security—for instance, during chest infections—which led to delays in seeking medical attention, or possibly because it caused diuretic atrophy of the respiratory muscles through “resting” them at night.

In practice, therefore, non-invasive ventilation is delayed until respiratory failure has developed and either the patient is symptomatic, has developed complications such as right heart failure or polycythaemia, or is considered to be at risk of premature death from respiratory failure. The decision when to initiate treatment, usually at night only, may be straightforward—for instance, in childhood proximal spinal muscular atrophy, previous poliomyelitis, slowly progressive myopathies, or muscular dystrophies without bulbar involvement. In other situations the decision may not be so clear cut particularly if the neurological disabilities are diverse as in, for instance, multiple sclerosis and multiple system atrophy. Motor neuron disease is the best example and exemplifies how ventilation can, if correctly used, bring dramatic symptomatic benefit but if indiscriminately used can simply be an extra physical and psychological burden for the patient and family. The best results are obtained if the condition is not progressing rapidly, if the non-respiratory disability is not too extensive, if bulbar function is relatively preserved, and if the diaphragm has been selectively paralysed.30-32 The success of treatment is also determined by the patient’s motivation to maintain an independent life and to cope with the successive problems that motor neuron disease presents.33 Clinical assessment of these important but ill defined attributes is essential when selecting patients with complex neurological disorders for home ventilation.

Most patients do not become totally independent even when they return to their home life and the presence, attitudes, and capabilities of their family and other carers are important to assess. Much depends, however, on the hospital’s preparation for discharge. The initiation of ventilatory support involves educating the patient and family, anticipating practical difficulties with the ventilator in the home, and encouraging an attitude of independence while the hospital retains a supportive role for both the patient and family. Careful liaison with the community services, planned readmissions to assess the patient’s progress, and the ability to rapidly repair or replace equipment that may fail are all important factors in building confidence and providing continuity of care.34 Does all this improve the patient’s quality of life? The answer that the patients give is often different from that of the health care professionals.35 Their values and goals may be quite different from those of the hospital team who, in general, tend to overestimate the importance of physical abilities to the patient and underestimate the value of the patient’s self respect, control of their environment, and social contacts.36 Most patients who require nocturnal ventilation adapt to their physical limitations and develop coping strategies that enable them to maintain their quality of life.27 Why then has long term ventilatory support not been used as much in neurological disorders in the United Kingdom as in other countries? In France and the United States, for example, at least twice as many patients are treated with home ventilation. The ventilators cost around £3000 but their maintenance is cheap and they should remain in service for at least five years. Cost alone is, therefore, probably not the problem. The difficulty is in assessing who is likely to benefit from treatment and, particularly in the more complex situations, in providing the postdischarge service to maintain the patient’s independence in the home. The experience and resources required for accurately selecting who should have treatment and for providing the required level of care is concentrated in specialist centres, but despite this home ventilation should be carefully considered whenever chronic respiratory muscle weakness becomes clinically significant. J M SHINEERSON

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