Lesson of the Week

Motor neurone disease presenting as respiratory failure

B AL-SHAikh, W KINNEAR, T W HIGENBOTTAM, H S SMITH, J M SHNEERSON, I WILKINSON

Diaphragmatic weakness may be the presenting feature of motor neurone disease, though the diagnosis may be difficult in the presence of chronic lung disease. We report a patient in whom the resulting hypoventilation at night had passed unrecognised.

Case report

A 65 year old man was referred to hospital for psychiatric assessment. After retiring six months earlier he had become confused and irritable. His wife described hypersomnia and had observed that his sleep was disturbed by regular waking. He was not obese and had stopped smoking 20 years previously. A left pleurodesis had been performed 10 years previously for recurrent pneumothoraces, the only recent respiratory history being of recurrent attacks of winter bronchitis. The only abnormality found on examination was impaired intellectual function. The chest radiograph showed pleural thickening on the left side. An isotope brain scan was normal, and the patient was discharged from hospital with a diagnosis of presenile dementia.

He was readmitted as an emergency four weeks later. On examination he was confused and cyanosed, but the only abnormal respiratory sign noted was rapid shallow breathing. There were no focal neurological signs. Arterial blood gas analysis while breathing air showed an arterial oxygen pressure (P_{aO_2}) of 3.8 kPa (28.5 mmHg) and an arterial carbon dioxide pressure (P_{aCO_2}) of 11.5 kPa (86.2 mmHg) with a hydrogen ion concentration of 45 nmol(nEq)/l. Because of the patient's hypoventilation a doxapram infusion was started, and the P_{aCO_2} fell to 7.5 kPa (56.2 mmHg). This improvement was maintained for 48 hours until he suddenly developed cardiorespiratory arrest. He was intubated and resuscitated, and on intermittent positive pressure ventilation with a fractional inspired oxygen concentration of 0.4 his arterial blood gases showed a P_{aO_2} of 16 kPa (120 mmHg) and P_{aCO_2} of 6.3 kPa (47.2 mmHg).

Despite a dramatic improvement in intellectual function attempts to wean the patient from ventilatory support after 48 hours were unsuccessful, and a tracheostomy was performed. Over the next two weeks he was gradually weaned from daytime ventilatory support and became fully independent during the day. He still required artificial ventilation by tracheostomy at night. He was fitted with a specially constructed cuirass respirator.¹ External negative pressure ventilation with this cuirass at night maintained normal oxygen saturations and transcutaneous carbon dioxide concentrations, and the tracheostomy was closed.

Careful neurological examination at this stage again failed to show any abnormality. The patient was orthopnoeic, and paradoxical abdominal motion was noted. The forced expiratory volume in one second (FEV_1) was

Diaphragmatic weakness and respiratory failure may be the first signs of motor neurone disease. Failure to recognise this may lead to cardiorespiratory arrest. External negative pressure assisted ventilation at night may enable such a patient to return home and lead an independent life

1.1 l with a forced vital capacity (FVC) of 1.9 l. The total lung capacity was 4.38 l (predicted 6.25 l) with a residual volume of 2.38 l (predicted 2.3 l). The peak expiratory flow rate was 150 l/minute and the maximal inspiratory pressure measured at the mouth with an occluded mouthpiece was 18 cm of water. Transdiaphragmatic pressure during sniffs was recorded in the sitting position with gastric and oesophageal balloons.² The maximal transdiaphragmatic pressure was 7 cm of water (normal range >80 cm of water), and he was unable to generate a positive intragastric pressure. The ventilatory response to carbon dioxide, assessed with a hyperoxic re-breathing method,³ was 9.1 l/kPa (68.2 l/mmHg) (normal range for our laboratory 8.3-26.4 l/kPa (62.5-197.8 mmHg)).

Two weeks later fasciculation of the tongue and legs was noted for the first time. A muscle biopsy examination was performed, which showed evidence of neurogenic atrophy, and motor neurone disease was diagnosed. The patient was taught to apply his cuirass himself. He was discharged from hospital and over the next eight months led an independent life in his own home. Although he developed dysarthria, he was able to resume his main hobby of gardening and perform maintenance work on his house. He spent an average of two days each month in hospital over the next eight months and died suddenly at home one night having got up to pass urine. Necropsy was not performed.

Discussion

Although respiratory insufficiency often develops in the late stages of motor neurone disease, presentation with respiratory features is uncommon. Patients presenting with acute respiratory failure due to motor neurone disease may initially be thought to have infection, airflow obstruction, or pulmonary oedema.⁴

Respiratory muscle weakness may be difficult to diagnose, particularly in patients with other respiratory diseases. Our patient had evidence of airflow obstruction, and this was thought to be his main problem when he presented in respiratory failure. At this time his hydrogen ion concentration was normal, suggesting that the high P_{aCO_2} reflected chronic hypoventilation. Maximal inspiratory pressure measurements are more reliable indicators of muscle power but depend on considerable cooperation on the part of the patient. In the measurement of transdiaphragmatic pressure a sniff may be easier to perform than static inspiratory efforts.⁴ The results of inspiratory muscle testing in our patient showed severe diaphragmatic weakness.

In the absence of specific pointers to respiratory disease hypoxia may be overlooked as a cause of impaired cerebral function, particularly when hypoventilation occurs mainly at night; in our

Intensive Care Unit, Hinchinbrooke Hospital, Huntingdon, Cambridgeshire

B AL-SHAikh, MB, senior house officer in anaesthetics
T W HIGENBOTTAM, MD, MRCP, consultant physician
H S SMITH, MB, FFRACS, consultant anaesthetist
I WILKINSON, MD, FRCP, consultant neurologist

Assisted Ventilation Unit, Newmarket Hospital, Newmarket, Suffolk

W KINNEAR, MA, MRCP, medical research registrar
J M SHNEERSON, MD, MRCP, consultant physician

Correspondence to: Dr T Higenbottam, Respiratory Physiology Laboratory, Papworth Hospital, Cambridge CB2 2QQ.

patient the abnormal sleep pattern was misinterpreted. Nocturnal hypoventilation occurs in patients with diaphragmatic paralysis,⁵ deformity of the chest wall,⁶ airflow obstruction,⁷ and disorders of the respiratory centres.⁸ Correction of this by assisted ventilation improves the quality of daytime life and may lead to prolonged survival. Even in patients with progressive neuromuscular disorders, such as motor neurone disease, the use of mechanical ventilatory support at home may enable the patient to enjoy a good quality of life for months or years.⁹ The use of a cuirass respirator at home in this patient allowed him to have eight months' good quality life. His family were aware of the poor prognosis but were grateful for the extra few months with him.

In conclusion, the possibility of progressive neuromuscular disease as an underlying cause in patients presenting with acute hypoventilation should not discourage the rapid establishment of adequate ventilation by artificial means.

MATERIA NON MEDICA

Zimmer to Alhambra

One of the most pleasant observations of a recent holiday abroad was the number of elderly and disabled people who had made their way to the sun—a far cry from my younger days when only the fit could make the journey. Most hotels overseas now attempt to meet the needs of the frail, those who have difficulty in walking, and people confined to wheelchairs. It is a completely new world.

On a trip to Grenada we had a lady with us who could not walk without the aid of a Zimmer frame. We all helped her on and off the coach and took it in turns to push the borrowed wheelchair when we reached the Alhambra. She saw as much of the sultan's palace as did the rest of us.

We were beaten only once. Lunch had been arranged at a nearby restaurant and the stairs to the dining room were rather steep. However, five young men came to our rescue. They were in costume: black knee breeches, a dark blouse, and a white rufflet around the neck. On their left breast was the symbol of Grenada, the three pomegranates. They were a group of singing guitarists who would later entertain us through lunch. Without more ado, they carried the lady up the stairs and, afterwards, took her down again.

During the recital they explained to the audience that they were poor medical students and this was their way of raising money for their fees. They were also selling cassettes of their music. I bought a couple, and play them still with pleasure, recalling a very happy day. I hope they succeed with their studies and make as good and caring doctors as they do young, happy, and helpful musical medical students.—WILLIAM THOMSON, chief administrative medical officer, Lanarkshire.

Two mountains, two peoples

The first mountain was called Chacaltaya. It is one of the glistening snowy peaks that form a sparkling tiara around the city of La Paz. I joined a group of rich Bolivians who were off for a day's skiing on the country's only ski slope. We had 600 feet to climb and our vehicle was an old snub nosed Mercedes bus. Roberto, our driver, was also old and snub nosed, but from a different maker. He, in His wisdom, had blessed Roberto with an abundant sense of humour but a disturbing absence of fear.

On Chacaltaya's lap we stopped while Roberto performed a little ceremony: he placed snow chains on the wheels and a crash helmet on his head. Thereafter the track was rocky, narrow, and precipitous. One hour later we were nearing the top when the bus went into a backward slide on a snowy bend. Only a few went over the edge: they were on the rear seat and their bottoms sat over a void. Roberto was left to laugh alone and we trudged the final distance on foot. I was soon left behind, trying to balance Broddingnagian breaths with slow staggering steps. It amused me that half my companions had not come to ski at all, merely to take the air. This seemed to typify their position of privilege. The air at 18 000 feet was a rare and valuable item.

Lunch was tortillas and omelettes washed down with maté de coca. My attempts at a siesta were thwarted by an alarming propensity to respire with a Cheyne-Stokes rhythm. Out on the piste the pace for some was necessarily slow, but the views were majestic. Around us roamed the ragged Andes,

References

- 1 Brown L, Kinnear WJM, Sargeant KA, Shneerson JM. Artificial ventilation by external negative pressure: a method for manufacturing cuirass shells. *Physiotherapy* 1985;71:181-3.
- 2 Miller JM, Moxham J, Green M. Maximal sniff in the assessment of diaphragm function in man. *Clin Sci* 1985;69:91-6.
- 3 Read DJC. A clinical method for assessing ventilatory response to carbon dioxide. *Aust NZ J Med* 1967;16:20-32.
- 4 Miller RD, Mulder DW, Ward S, *et al*. Exertional dyspnoea: a primary complaint in unusual cases of muscular atrophy and amyotrophic lateral sclerosis. *Ann Intern Med* 1957;46:119-25.
- 5 Skatrud J, Iber C, McHush W. Determinants of hypoventilation during wakefulness and sleep in diaphragmatic paralysis. *Am Rev Respir Dis* 1980;121:587-93.
- 6 Mezon B, West P, Israels J. Sleep breathing abnormalities in kyphoscoliosis. *Am Rev Respir Dis* 1980;122:617-21.
- 7 Stradling J, Lane D. Nocturnal hypoxaemia in chronic obstructive pulmonary disease. *Clin Sci* 1983;64:213-22.
- 8 Guilleminault C, Cumiskey J, Dement W. Sleep apnoea syndrome: recent advances. *Adv Intern Med* 1980;26:347-72.
- 9 Sivak E, Streib E. Management of hypoventilation in motor neurone disease presenting with respiratory insufficiency. *Ann Neurol* 1980;7:188-91.

(Accepted 4 March 1986)

below lay the barren altiplano and miniaturised La Paz; beyond glinted the vast mirror of Lake Titicaca. The vistas were brief, cut short by cumulus. There was a long dull roar, and we were wrapped about by thunder clouds. My hair began to writhe and crackle under its woolly hat. Chacaltaya had been an electrifying and breathtaking experience.

One week later and 40 miles further south I was donning a miner's hat at the restful altitude of 13 000 feet. This was Potosi and a mountain that had to be seen from the inside. The Spaniards first began digging here 450 years ago. Initially it was for massive quantities of silver that financed wars in Europe. Tin is now the main mineral extracted but working conditions have improved little. I spoke with the "medico" in the mine. There are many injuries; trauma and miners' lung ensure that few work past their fortieth birthday. Since the collapse of the International Tin Council they had not been paid. The government miners were best off as they received free handouts of bread and coca leaves. These they chewed throughout the day to numb the sense of hunger and cold.

Inside, the miners worked in groups of four. They waved cheerily, calling out, "Buenos tardes! Loco gringo!" I was mad to enter their Stygian world. The pungent air left a bitter taste. The dark walls glittered with fool's gold. Bright cupreous blue stalactites dripped a corrosive acid that could put out an eye and etched the pit props. Chasms yawned in the roof where the vertical seams had been worked. Whichever way I looked at it there was a wealth of difference between their world and mine. But the real tragedy was the difference I had seen in the one country on the two mountains.—WILLIAM CLAYTON, London.

What is seasonal affective disorder?

The term "affective disorder" is usually employed for the common depressive illness (endogenous depression) and for the less common mania. In some patients these illnesses recur over many years. Even if the timing of recurrences were random, within a large cohort of such patients several would inevitably have begun their last few illnesses during, say, the winter. The term "seasonal affective disorder" has been used to hypothesise that in patients who have had recurrences during one season of the year the relation with the seasons is not chance but causal. A causal relation, however, remains to be demonstrated. We have inherent biological rhythms, and many species possess, independently of such things as day length, rhythms that make the animals less energetic and relatively unresponsive during the winter, when food for energy requirements is scarce. Such a rhythm has not yet been shown for man. We speak of gloomy winter days and of gloomy and black thoughts in depression; some writers assume that patients may present with feelings of depression because of less light in winter, and they recommend gazing at banks of electric lamps. Adequate experimental controls are impossible here, and feelings of depression do not lend themselves to precise measurement. Any value of treatment by electric light should be attributed to the power of suggestion, even though many of us do feel uplifted in our spirits on days of spring sunshine.—IAN OSWALD, professor of psychiatry, Edinburgh.