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Motor neuron disease presenting with acute hypercapnic respiratory failure

TO THE EDITOR: Motor neuron disease (MND) is rare, and respiratory failure at initial presentation is even rarer. Most patients present with asymmetrical limb weakness.[1] We present a case of MND presenting with acute hypercapnic respiratory failure.

A previously well 61-year-old black African man who worked as a trauma nurse complained of breathlessness while at work. He reported a 3-month history of intermittent shortness of breath and a 2-week history of a non-productive cough. He was initially assessed as stable with normal vital signs. However, 45 minutes later while in the emergency department, he became tachypnoeic, hypoxic and confused. Arterial blood gas (ABG) measurement (Table 1) demonstrated acute hypercapnic respiratory failure. He was initially started on facemask oxygen and subsequently escalated to continuous positive-pressure ventilation, resulting in an improvement clinically and on repeat ABG measurement (Table 1).

The initial differential diagnosis included a pulmonary embolus and an infective process (including COVID-19). However, further investigations for these conditions were negative.

A more detailed history revealed motor deficits and fasciculations. The patient reported a 3-month history of difficulty opening glass vials of medication when at work, suggestive of intrinsic hand muscle weakness, and a 2-month history of 'jumping' pectoral muscles. He was also intermittently confused on awakening, which was suggestive of carbon dioxide narcosis, with relative hypoventilation during sleep. He had unintentional weight loss of 25 kg of muscle bulk over the last few months.

On examination, the patient was in respiratory distress and globally wasted with florid shoulder girdle fasciculations. The cranial nerves were normal. There was no nystagmus and no ophthalmoplegia. Bulbar signs were not elicited, and there was a normal jaw jerk. There was no extension or neck flexion weakness. On motor examination, he had proximal and distal muscle wasting of the upper limbs with split hand wasting and fasciculations. He had normal tone with reduced power of 4/5 in all muscle groups and brisk reflexes of 3/4. His lower limbs had no obvious wasting, normal tone, and reduced power proximally and preserved power distally. He had brisk knee reflexes of 3/4 with no ankle jerks and an upgoing plantar reflex on the right. The findings on sensory examination were normal, and he was not ataxic. He was assessed as having a predominant or pure motor syndrome with combined upper and lower motor neuron signs.

Table 1. Serial blood gas levels and laboratory results

Variable	Initial (reference)	After non-invasive ventilation
pH	7.23 (7.35 - 7.45)	7.34
PO_2 (mmHg)	50.9 (80 - 100)	185
PCO_2 (mmHg)	93.4 (35 - 45)	57
H_2CO_3 (mEq/L)	29.9 (22 - 26)	32
D-dimers (mg/L)	0.13 (<0.50)	n/a
C-reactive protein (mg/L)	2 (0 - 10)	n/a
Procalcitonin (ng/mL)	0.04 (<0.05)	n/a

PO_2 = partial pressure of oxygen; PCO_2 = partial pressure of carbon dioxide; H_2CO_3 = bicarbonate; n/a = not applicable.

Electrodiagnostic studies were performed. Nerve conduction studies showed normal latency and conduction velocity, with decreased amplitude in motor fibres. Sensory fibres were normal. Repetitive nerve stimulation of the accessory nerve demonstrated no decrease in the amplitude of the motor response. The patient was diagnosed with MND, amyotrophic lateral sclerosis variant. He had an atypical presentation with acute hypercapnic respiratory failure.

Once normocarbic, the patient was slowly weaned off non-invasive ventilation with arrangements to continue bilevel positive airway pressure ventilation at home. However, he experienced two significant decompensations. At his request he was not reintubated, and he subsequently died. His survival time from presentation to death was 5 weeks.

MND, also known as Lou Gehrig disease and amyotrophic lateral sclerosis, is a degenerative disease that involves both upper and lower motor neurons. Clinical presentation depends on the initial affected body segment and may manifest as upper motor neuron- or lower motor neuron-type pathology.[1] While the El Escorial criteria are more well known, the current standard of diagnosis is based on the updated Gold Coast criteria, which allow for earlier diagnosis, earlier intervention, and enrolment into clinical trials.[1]

Predominant and pure respiratory muscle weakness is an uncommon initial presentation of MND, with an estimated frequency between 2.7% and 5.0%.[2,3] These patients experience dyspnoea or orthopnoea, voice changes, a weak cough, and features of carbon dioxide retention including morning headaches, hallucinations, confusion and daytime somnolence.[3] Even within this subset, acute type 2 respiratory failure, as seen in this case, is rare.

Despite our patient's preceding asymmetrical limb weakness, he worked as a nurse until his acute deterioration. This aspect of the case highlights patients' ability to functionally compensate for illness and the importance of a detailed clinical assessment.

Most patients with MND die within 3 - 5 years of diagnosis.[1] Older age at onset, rapid change in function, executive dysfunction, respiratory subtype MND or an increased level of the biomarker neurofilament light chain are associated with decreased survival.[4] Our patient had rapid functional decline and respiratory weakness.

Most treatment is symptom directed. The two main disease-modifying drugs are riluzole and edaravone. Riluzole, which improves survival, decreases glutamate-induced excitotoxicity.[5] Edaravone decreases the rate of functional decline by acting as a free radical scavenger and decreasing oxidative stress.[6] In our setting, a public hospital in a middle-income country, availability of these drugs is limited. Research on emerging therapies such as gene therapy offers promise.

In conclusion, type 2 respiratory failure is easily diagnosable on ABG measurement. Owing to its various possible causes, a structured approach to diagnosis is essential. The presentation pattern of MND is broad and, as in this rare case, includes acute respiratory failure.

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