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Case Report

The Recognition of Respiratory Failure in Amyotrophic Lateral Sclerosis - Delays and Distress

Oliver D[1]*, Banerjee S[2] and Vincent-Smith L[2]
[1]Tizard Centre, University of Kent, UK
[2]Respiratory and Sleep Medicine, Medway Maritime Hospital, UK

***Corresponding author:** David Oliver, Tizard Centre, Cornwallis North East, University of Kent, Canterbury, UK

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Abstract

Patients may initially be seen with respiratory symptoms when first presenting with amyotrophic lateral sclerosis/motor neurone disease. These cases show how assumptions can be made regarding these symptoms leading to misdiagnosis and delays in the diagnosis of ALS/MND. A full clinical assessment of patients is essential in ensuring these delays are avoided so that patients may receive the appropriate care.

Keywords: Motor neurone disease; Respiratory symptoms; Delay in diagnosis; Clinical assessment; Amyotrophic lateral sclerosis

Introduction

Amyotrophic Lateral Sclerosis/Motor Neurone Disease (ALS / MND) is a progressive disease of the nervous system affecting primarily, but not exclusively, motor neurones. The cause is unknown, but in 5-10% of cases there is a family history and there is increasing evidence of a genetic basis for the disease, with an unknown provoking environmental or other genetic involvement [1]. Most people with ALS/MND present with muscle weakness, in arms or legs, or as swallowing or speech problems due to bulbar involvement. However a small proportion of people present with respiratory muscle weakness [2] often as an emergency when they may even need ventilation, by non-invasive ventilation or invasive ventilation with a tracheostomy.

Case Presentation

These cases show the issues that may arise when there is a misdiagnosis of symptoms, without a full assessment of the person's history, symptoms and signs. Increasingly patients may be seen as presenting with a particular symptom complex which is considered to be related to a particular diagnosis, without consideration of the whole patient and a careful assessment of the situation. The names and details have been changed to protect anonymity.

Case 1

Mr M was a 70 year old man who had suffered from heart failure since 1999 when he had undergone a coronary arterial bypass graft. He had been seen on a regular basis by cardiology services, including the specialist heart failure team. In September 2011 he was seen with increasing dyspnoea, which he stated had been present for over a year. His main symptom of waking at night was thought to be paroxysmal nocturnal dyspnoea. At the end of January 2012 he was admitted to hospital with shortness of breath on minimal exertion and was found to be in fast atrial fibrillation. This was treated but the episodes continued.

In March 2012 he underwent an angiogram but developed both local infection and septicaemia. A pseudo-aneurysm developed in the groin which required a thrombin injection but he deteriorated and became confused. He was found to have an elevated carbon dioxide level and non-invasive ventilation was started, and a diagnosis of Type 2 respiratory failure was made. A flat diaphragm was noted on X ray. In early May, two months after admission to hospital, a neurological examination as performed for the first time. Muscle fasciculations were noted and a neurological opinion was requested. However he required an above knee amputation as the infection was spreading and he was very ill following this.

In early July the diagnosis was confirmed as ALS/MND and he was told this later that month. He was very keen to return home and with support from the physiotherapist, occupational therapist and the palliative care team he was able to return to a specially rented flat where he died three weeks later.

Case 2

Mrs L was an 82 year old lady. She had suffered with angina for several years and had hypertension and hyperlipidaemia. In November 2010 she had been seen for unexplained weight loss by an elderly care physician, and no cause was found but no neurological examination is recorded. In June 2012 she was admitted to hospital with dizziness and "near to collapse" and was found to have atrial fibrillation. In October she was seen by a physician and was noted to have a drooping head, shaking hands and was short of breath. Myasthenia gravis was suggested and a referral was made to a respiratory physician. However in November she collapsed at home and was admitted again and was noted to have a postural hypotension, light headedness on standing and weight loss. She had a possible seizure on the day after admission and blood gases were found to show hypoxia (pO_2 5.19 kPa) and hypercapnic (pCO_2 14.3 kPa). A neurological examination was performed and she was noted to have small muscle wasting in her hands and her swallowing was reduced. A speech and language therapist noted a "tremulous tongue". Non-invasive ventilation was started and a diagnosis of "anterior horn cell disease" was recorded. The diagnosis of ALS/MND was suggested by a neurological opinion and this was confirmed by a consultant neurologist in early December. She was seen by the Hospital Palliative Care Team and then transferred to the hospice. She deteriorated quickly and died 4 days later.

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Discussion

These cases show how the incipient respiratory failure due to ALS/MND can be missed and confused with other pathological processes. However in both cases the patients had complained of symptoms that could have indicated respiratory muscle weakness but due to there being another diagnosis these were missed and assumed to be related to the previous pathology. There would seem to be the loss of clinical acumen and consideration of the diagnosis of the symptom. The patient's symptoms were "assumed" to be due to the pre-existing diagnosis, and this limited the assessment. Both patients did give a history of increasing dyspnoea, orthopnoea and poor sleep but these symptoms were not elicited on admission, and once the diagnosis was "made" there was no further careful history taking or consideration of alternatives. Moreover there was over reliance on investigations - both had "normal " X rays, although in retrospect the diaphragm was noted to be elevated, and there was limited clinical examination of the patient [3]. In Mr M's case he had been admitted with breathlessness and weakness but no neurological examination is recorded for two months and in Mrs L's case no neurological examination was performed despite an admission with dizziness or following a collapse.

In the past the triad of history taking, examination and then investigation was the normality and taught to all medical students. However over time there has been increasing reliance on investigation and the overall holistic assessment has been lost. Within palliative medicine the importance of listening to the patient and carefully assessing the cause of symptoms has been stressed and the "impeccable assessment" is the mainstay of the World Health Organisation's definition of palliative care [4]. It is sad to see that patients can be misdiagnosed for long periods of time and the management of their symptoms and problems are missed. Hopefully the increasing pressure on a more holistic and caring approach to patients within hospital, following the Francis Report into the deaths within a UK hospital may also establish the need for the basics of medicine-"listen and look at the patient before all else" [5].

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