CASE REPORT

Motor neurone disease presenting as polycythaemia

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SUMMARY

Motor neurone disease (MND) is a chronic, progressive and currently incurable neurodegenerative disorder. This case report discusses an instance of MND presenting initially as polycythaemia, caused via insidious respiratory failure through ventilatory insufficiency. This case aims to improve clinicians' awareness of this atypical presentation and highlights the need for a high index of suspicion of respiratory failure in any patient with polycythaemia. Finally it demonstrates an improvement in quality of life associated with the use of non-invasive ventilation (NIV) in a patient with MND.

BACKGROUND

Motor neurone disease (MND) can pose a diagnostic challenge as a result of its varied presenting symptoms and subsequently wide differential diagnosis. This case report reminds clinicians that it can often be overlooked as a diagnosis in the absence of obvious neurological symptoms.

An instance of polycythaemia preceding the onset of MND is presented here. This rare presentation is not represented, to the best of our knowledge, within published literature to date and the overall case highlights the insidious nature with which MND can present.

CASE PRESENTATION

A 59-year-old security officer was initially referred to haematologists by his general practitioner for investigation of polycythaemia and weight loss after presenting with reduced exercise tolerance and weight loss.

Relevant medical history included hypertension, hypercholesterolaemia, mild emphysema and testicular cancer cured after orchidectomy and adjuvant chemotherapy without the need for radiotherapy 30 years previously. The patient, a smoker with a 40-pack-year history, lived independently with his wife and had no significant family history of note.

INVESTIGATIONS

Haematological investigations revealed low erythropoietin levels but negative JAK2 (exons 12 and 14) coupled with a normal bone marrow aspirate, suggesting secondary polycythaemia (Hb 19.2 g/dL). Radiological investigations including CT of the thorax, oesophagogastroduodenoscopy and flexible sigmoidoscopy were unremarkable except for a mild ulcer treated with omeprazole and confirmed to have been eradicated by repeat endoscopy.

Six months following initial haematological referral, urgent respiratory review was requested when the patient attended haematology follow-up with

increasing neck pain, continued weight loss and dyspnoea in the absence of a cough or sputum production.

Prior to respiratory review, the patient was admitted as an emergency with impaired consciousness and acute on chronic hypercapnic respiratory failure requiring immediate invasive mechanical ventilation (IMV) (ABG on admission: pH 7.14, pO₂ 8.12 kPa, pCO₂ 18.4 kPa, HCO₃⁻ 37.2 mmol/L).

During this admission, he was noted to have rapidly progressing thoracic kyphosis and a loss of cervical lordosis with reduced sniff nasal inspiratory pressures of 15 cm H₂O. No abnormal neurology was identified. Subsequent spirometry revealed a restrictive pattern (FEV₁ 1.36 L (52% predicted), FVC 1.74 L (54% predicted), FER 78%).

The patient required NIV during respiratory weaning from IMV and prior to discharge had to be started on domiciliary NIV before being referred for neurological review regarding his worsening thoracic kyphosis and loss of cervical lordosis (figure 1) in the presence of respiratory failure. Attempts to obtain MRI cervical and thoracic spine images during admission were unsuccessful due to breathing problems preventing adequate positioning.

By the time of neurological review 3 months later and just over a year from initial haematological referral, the patient remained able to mobilise unaided. However, he had developed significant proximal and distal muscle wasting and noticeable quadriceps fasciculations. Examination revealed global weakness affecting all limbs proximally and distally, slightly more pronounced on the right hand side. Cranial nerve and sensory functions were intact with speech unaffected and an absence

Figure 1 X-ray demonstrating loss of normal cervical spine lordosis.

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of tongue fasciculation or wasting. Reflexes were brisk, plantars downgoing, coordination intact and Hoffman's sign could not be elicited. Subsequent electromyography and nerve conduction studies supported a diagnosis of MND.

DIFFERENTIAL DIAGNOSIS

- ▶ Chronic respiratory failure due to MND-induced left hemidiaphragm paralysis resulting in compensatory polycythaemia.
- ▶ Paralysed left hemidiaphragm due to phrenic nerve palsy from cervical myelopathy.

TREATMENT

Domiciliary NIV and MND multidisciplinary services input.

OUTCOME AND FOLLOW-UP

Survived for 8 months on domiciliary nocturnal NIV.

DISCUSSION

Timely diagnosis is vital to enable the complex multidisciplinary care planning necessary to maximise patients' quality of life during the latter stages of MND.¹ Yet, the absence of a definitive diagnostic test, multimodal presentation with an extensive differential diagnosis and insidious onset, continue to cause significant diagnostic delays.¹⁻³ MND presenting to a haematology clinic as secondary polycythaemia is not reported to the best of our knowledge within literature to date.

Presentations of MND besides motor weakness symptoms are largely respiratory in nature, including dyspnoea⁴ and acute respiratory failure requiring NIV.^{1 5} This case report has described how MND onset symptoms can also be haematological in nature: in this case the patient initially presented with polycythaemia. The importance of assessing for respiratory failure in all patients with polycythaemia is reinforced as well as the need to consider insidious respiratory failure as the cause of secondary polycythaemia cases.

NIV is known to improve symptoms, quality of life and survival among patients with MND by combating the significant morbidity and mortality associated with respiratory muscle weakness. Guidelines suggest it should be considered for patients with symptoms and/or signs of respiratory failure coupled with impaired respiratory function tests.⁶ Recent years have seen a significant rise in its use of 3.4-fold between 2000 and 2012.⁷ This likely represents greater awareness of its role among clinicians coupled with clear guidelines regarding indications and methods

of use within this complex patient group. Our case report highlights the ability of NIV to extend patient survival by several months.

Learning points

- ▶ Motor neurone disease (MND) can present atypically and a high index of suspicion is required to achieve early diagnosis.
- ▶ Insidious respiratory failure should be seen as a cause of polycythaemia.
- ▶ Neuromuscular diseases must be actively excluded when treating respiratory failure of unknown aetiology.
- ▶ Non-invasive ventilation (NIV) should be considered in patients with symptoms/signs of respiratory failure and impaired respiratory function tests.
- ▶ Quality of life of patients with MND can be improved with early diagnosis, multidisciplinary input and NIV initiation.⁶

Contributors NS-V reviewed the patient's notes, obtained patient consent and was responsible for drafting and revising the case report. RM initiated the project and is the guarantor. RM and JB assisted with drafting and revising the case report. KM provided patient care and has reviewed the case report.

Competing interests None.

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