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CASE REPORT

Dystrophia Myotonica with Hypercapnic Respiratory Failure – Local Challenges and Ethical Dilemmas

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ABSTRACT

Dystrophia myotonica type 1 (DM1) is an adult-onset progressive form of muscular dystrophy which eventually lead to respiratory failure. Non-invasive home mechanical ventilation (HMV) for these patients could result in a reduced risk of morbidity and mortality. The following case report describes the management of respiratory failure in a patient with DM, and the challenges that come with it especially in our Malaysian settings.

KEYWORDS: Dystrophia myotonica, respiratory failure, non-invasive, home mechanical ventilation

INTRODUCTION

Dystrophia myotonica type 1 (DM1) is an adult-onset progressive form of muscular dystrophy [1]. The disease prevalence ranges from 0.5 to 18.1 per 100,000 people [1]. Due to its progressive nature, respiratory failure is commonly the end point among the patients. This is due to the complex nature of the disease involving respiratory muscle weakness, reduced central respiratory drive, decreased chest wall compliance, and upper airway obstruction. Therefore, in certain situations, we can implement invasive or non-invasive home mechanical ventilation (HMV) for the patients, which could result in a reduced risk of morbidity and mortality. Due to the rarity of the disease and lack of good clinical studies, it is hard to have a one-size-fits-all management for each DM patient.

The following case report describes the management of a patient with DM, and the challenges that come with it.

CASE PRESENTATION

A 36-year-old gentleman presented with choking and coughing episodes after meals. He was diagnosed with dystrophia myotonica in 2003 at the age of 19 years. Previously, he had multiple ward admissions for lung infections in different hospitals. In 2018, he had severe pneumonia that required invasive mechanical ventilation and tracheostomy due to difficulty in weaning ventilation. He was discharged after four months of hospital admission with a bilevel positive airway pressure (BiPAP) home ventilator via tracheostomy. Unfortunately, he did not use the home

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ventilator and defaulted the follow-ups upon discharge. For his baseline functional status, he was able to mobilise using a wheelchair by himself and only required the assistance of one person for transfer.

On examination, he was febrile, and his oxygen saturation was 80% on room air via tracheostomy. He had frontal baldness, bilateral partial eye ptosis, wasting of the temporalis and facial muscles, and positive percussion myotonia of the tongue and thenar muscles. Reduced breath sound was noted over the right lung, and chest radiograph revealed collapse consolidation over the right upper zone (Figure 1). Blood tests, including gas analysis showed leucocytosis, elevated C-reactive protein and type 2 respiratory failure (Table 1). He was diagnosed with aspiration pneumonia.

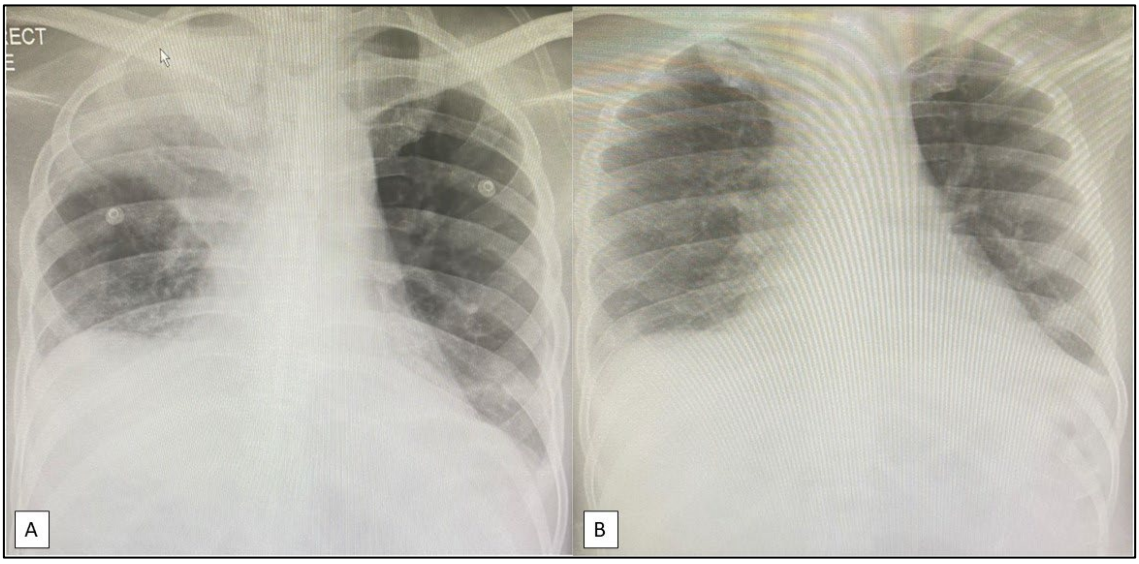

Figure 1 Chest radiograph during hospital admission. (A) Chest radiograph at presentation showed right upper zone collapsed consolidation and elevated right hemidiaphragm. (B) Repeat chest radiograph after 2-week of antibiotic, chest physiotherapy and non-invasive ventilation which showed improvement of the right upper zone collapse consolidation.

Table 1: Blood investigations at presentation

	Value	**Reference Range**
Full blood count		
Haemoglobin	16.4	12-15 g/dL
White Cell Count	16.6	4-10 x 10^9/L
Platelet	470	150-410 x 10^9/L
Hematocrits	52.4	36-46%
Renal profiles		
Urea	1.9	2.5-6.7 mmol/L
Sodium	139	136-145 mmol/L
Potassium	5.4	3.5-5.1 mmol/L
Arterial blood gases		
pH	7.37	
Partial oxygen	46.6	mmHg
Partial carbon dioxide	47.6	mmHg
Bicarbonate	27.8	mmol/L
Oxygen saturation	80%	
CRP	79	<0 mg/dl

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He was treated with intravenous piperacillin-tazobactam and received continuous positive airway pressure (CPAP) ventilation via tracheostomy. He improved after receiving two weeks of antibiotic and chest physiotherapy. Cultures of his sputum and blood were negative, and his chest radiograph showed a resolution of the right upper collapse and consolidation. Unfortunately, he had persistent elevated transcutaneous carbon dioxide (TCO_2) levels, which ranged from 54 to 60 mmHg despite good oxygenation (Table 2). He then received BiPAP ventilation with the aim of normalising his TCO_2. At the end of the third week of hospitalisation, his TCO_2 level was normalised at 44 mmHg with inspiratory positive airway pressure (IPAP) of 18 mmHg and expiratory positive airway pressure (EPAP) of 6 mmHg with a back-up respiratory rate of 16/min. The patient did not require supplemental oxygen at this point.

We re-discussed the option of a home ventilator with his family members, to which both the patient and family members agreed. During the hospital admission, the family members were re-trained on the home ventilator care and settings. In addition, the family members were taught on nasogastric tube feeding and airway clearance technique. Finally, we re-emphasised the importance of regular follow-ups, particularly the indications and settings of the home ventilator, which differed as the pneumonia resolved and the disease progressed. He was discharged with the home ventilator after 21 days of admission.

Table 2 Arterial blood gases (ABG), oxygen saturation and non-invsive ventilator setting

ABG Trend	On admission	Day 1	Day 2	Day 3 (am)	Day 3 (pm)	Day 5-18	Prior discharge
pH	7.37	7.31	7.33	7.3	7.36		7.46
PaCO2	47.6	58	54.8	60.7	54.9	50-55*	44.6
PaO2	46.6	95.3	120.8	95.4	103.4		97.2
HCO3	27.8	30	28.9	32	31.5		31
SpO2	80%	100%	100%	100%	100%		98%
Ventilator setting & supplementary oxygen		CPAP, FiO2 0.3, PEEP 8	Tachymask 5L/min	Tachymask 5L/min	CPAP, FiO2 0.3, PEEP 8		BiPAP, IPAP 18, EPAP 6, ST 16

* Transcutaneous carbon dioxide

DISCUSSION

Dystrophia myotonica (DM) is an autosomal dominant disorder with a reduced life expectancy of about 54 years [1]. Majority of the patients succumb to the disorder due to respiratory failure [1], which is driven by complex interactions between respiratory muscle weakness [2], reduced central respiratory drive [3], decreased chest wall compliance [4], and sleep apnoea [5]. Home mechanical ventilation has been utilised in DM patients with chronic hypercapnic respiratory failure, which has been shown to lead to improvement of gas exchange [6] and survival [7, 8]. However, in the present case, the HMV was a complex treatment decision due to the overwhelming responsibility for the care of the patient on his caregivers.

The decision to initiate the HMV for this patient was based on the hypercapnic respiratory failure evidenced by the elevated carbon dioxide >45 mmHg. Other indications for HMV are reduction of 20% or 500 mL of forced vital capacity in sitting and supine position, maximum inspiratory pressure of less than −60 cm H_20, maximum expiratory pressure of less than 40 cm H_2O, peak expiratory flow of less than 270 mL, and polysomnography with apnoea-hypopnoea index of >5/h [9]. Unfortunately, in the present case, the patient presented with acute respiratory failure and pre-existing tracheostomy. Majority of the respiratory assessments,

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including lung function tests and polysomnography, could therefore not be performed. Ideally, these assessments should be performed every 6 months in a multidisciplinary team clinic.

DM patients who were initiated with HMV following acute respiratory failure were shown to have a poor compliance post-discharge [10]. The initial assessment of hypercapnic respiratory failure following the pneumonia was questionable. It was unclear if this patient indeed had chronic respiratory failure, although the present findings of persistent elevated serum HCO_3 level supported this diagnosis. He might recover and become non-compliant as the indications of HMV had fully ceased. Another reason could be that the patient and carers were not well-informed regarding the aim of the HMV, which was normalisation of carbon dioxide and not improvement of oxygen saturation. Initially, both patient and carers were reluctant for HMV as the oxygen saturation was normal under room air. We were, however, able to convince both of them on the need of HMV to normalise carbon dioxide, which could help prolong his survival [7, 8].

Oxygen therapy is rarely needed in the setting of HMV as the underlying disease does not cause any form of lung parenchyma damage. The patient in the present report required oxygen only during his initial stay due to the infection, which cleared quickly after the course of antibiotic treatment and with the aid of physiotherapy for airway clearance. Despite these interventions, his hypercapnia persisted because of hypoventilation due to the ineffective or low tidal volumes and respiratory rate. Only bilevel positive airway pressure (BiPap) ventilation would address both the hypoxaemia and hypercapnia. In addition, a compulsory back-up respiratory rate in the ventilator setting was important to address the potential central apnoea or respiratory drive impairment [9].

It is pertinent to evaluate patients with neuromuscular diseases as they are likely to have facial and bulbar weaknesses. A facial morphology assessment allows us to select suitable mask interfaces. Patients with facial weakness are suited to oronasal masks or nasal interfaces with chinstraps to prevent excessive leaks [9]. On the other hand, patients with bulbar dysfunction may need a tracheostomy ventilation as it reduces the risk of aspiration [11]. During the inpatient ventilator adjustments for the patient in this case report, we included an adaptor with a leak port connected to a single limb connecting tube. The purpose of this contraption was to allow intentional air-leak for the washout of carbon dioxide.

Another important aspect to ensure compliance to HMV is to familiarise the patient and the carers with home ventilator care and setting through a supervised training [10]. We managed to address these while the patient was hospitalised over a few weeks. Similarly, the ventilator setting that aimed for carbon dioxide normalisation was done over a few days after the pneumonia had resolved. This arrangement allowed the patient to acclimatise to HMV and the appropriate setting to normalise carbon dioxide. Home ventilator vendor was contacted, and the existing home ventilator was serviced. This collaborative partnership also extended beyond the hospital admission as their roles were also incorporated into domiciliary care team once the patient was discharged.

The establishment of HMV within Asia-Pacific countries is rather limited due to the lack of expertise and funding. As HMV care consists of a large group of specialties extending their care to the community, it would also be labour-intensive. Malaysia has a publicly funded healthcare system, where the resources used in one area may affect negatively on the other groups of patients. Therefore, incorporating HMV services within our hospitals and community will require a huge amount of effort from many organisations. Our patient was fortunate enough to have the financial means of purchasing the BiPAP machine.

It is undoubtedly a difficult ethical decision to make in the management of patients with end-stage respiratory failure especially when the therapy choices become a question of life or death. The prognosis in DM patients is poor, and respiratory complications hasten the patient's demise if the life-sustaining mechanical ventilation is not implemented. Tracheostomy may be able to extend the patient's life, but without the guarantee of a good quality life. Therefore, it is essential to recognise the patient's autonomy over his own well-being in the decision of HMV during the discussion of long-term care. In fact, a study done on Motor Neuron Disease (MND) patients on HMV showed that the majority of caregivers considered that the best thing

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about ventilation was keeping the patients alive [12]. However, caregivers living with HMV-dependent individuals have a significant burden in the form of psychological and physical consequences [13]. These can inadvertently affect the quality of care given to the patients. We must never neglect the welfare of the caregivers during the discussion because after all they are the pillars of strength for the patients' care.

Summary of challenges and solutions for HMV in DM patients were as follows:

Challenges	Solutions
Hypercapnic respiratory failure	• Bi-level positive airway pressure ventilation (BiPAP) with compulsory back-up respiratory rate
Facial and bulbar weaknesses for suitable mask interfaces	• Nasal mask with the adjunct of chinstraps • Oronasal masks • Tracheostomy

CONCLUSION

Respiratory failure frequently occurs in DM patients as they progress in life. It is established that DM patients can be effectively palliated using HMV. This information can be discussed with the patients and their caregivers, allowing them enough time to consider the practical implications of the ventilatory support. The patients and their caregivers should be warned that this treatment can prolong survival but may be at the expense of their quality life. The patients' autonomy may vary but their dignity and integrity remain constant. Both physicians and patients should actively participate in the decision-making process so that the initiation of HMV can be planned around their daily activities. Clinical trials to measure the health economic aspects of HMV and its impact on quality of life are much encouraged especially in Malaysia.

Conflict of Interest

Authors declare none.

Authors' contribution

All authors reviewed and edited the manuscript and approved the final version of the manuscript.

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