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Decannulation in a pediatric patient with neuromuscular disease

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Abstract: Multiminicores disease is a hereditary neuromuscular disorder characterized by the presence of multiple 'nuclei' on muscle biopsy and clinical features of a congenital myopathy. The present case concerns a 10-year-old patient, diagnosed with multiminicores disease, tracheostomized since she was seven due to failed weaning and muscle weakness. The patient was referred to the Department of Cardio-respiratory Rehabilitation of the Clínicas Hospital from the National University of Asunción, presenting in her first evaluation chronic oxygen dependence (for more than 12 months), weak and non-functional cough with cough peak flow less than 160 L/m, chronic respiratory failure and hypercapnia (52 mmHg CO2ET). We did the follow-up in order to the Respiratory Rehabilitation guidelines contemplated in the Project for the evaluation, treatment and follow-up of patients with Neuromuscular Diseases, which was approved by the Superior Council of the Medical Sciences School from the National University of Asunción, these guidelines are based on scientific studies and publications done by Dr. John Bach (Rudgers University, Newart, New Jersey-USA) and his collaborating team from the Ibero-American Group for Respiratory Care in Neuromuscular Diseases. As a result, the patient was successfully decannulated, in an outpatient clinic, without hemodynamic decompensations, with excellent tolerance and without the requirement of hospital admissions.

Key words: neuromuscular disease; tracheotomy; respiratory rehabilitation; decannulation; multicores disease

1 Introduction

Multicores disease (MmD) is an inherited neuromuscular disorder characterized by the presence of multiple “cores” on muscle biopsy and clinical features of congenital myopathy. The marked clinical variability corresponds to genetic heterogeneity: the most easily recognizable classic phenotype is characterized by spinal stiffness, premature scoliosis, and respiratory involvement; it is caused by recessive mutations in the selenoprotein N gene (SEPN1), whereas recessive mutations in the skeletal muscle ryanodine receptor (RYR1) are associated with a wide range of clinical features [1].

Patients with congenital or acquired neuromuscular diseases (NMDs) experience progressive deterioration of respiratory function; if left untreated, this leads to high morbidity, with respiratory failure being the cause of death [2].

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Early detection of respiratory compromise and appropriate ventilatory support improve survival and quality of life in many patients [3].

Patients without NMD who are intubated are extubated based on a test of spontaneous breathing and ventilator weaning parameters. However, when intubated, patients with NMD do not meet these weaning parameters because they lack the necessary strength to breathe or cough; for this reason, they undergo tracheostomy [4,5].

The following is the clinical case of a pediatric patient diagnosed with MmD, who has had a tracheostomy for more than 3 years, is oxygen-dependent, and has chronic respiratory failure. She received treatment through the Project for the Evaluation, Treatment, and Follow-up of Patients with Neuromuscular Diseases, carried out by the Department of Cardiorespiratory Rehabilitation at the Hospital de Clínicas.

2 Case presentation

A 10-year-old school-aged girl with a medical diagnosis of multiminicore myopathy. Since she was 15 months old, she has had multiple hospitalizations due to respiratory exacerbations, most of which were treated with oxygen therapy and antibiotics. At age 7, she underwent a tracheostomy due to prolonged intubation and multiple failed weaning attempts. She was discharged on nocturnal oxygen therapy, and starting at age 8, she was prescribed nocturnal bi-level positive airway pressure (BIPAP) and continuous oxygen therapy. She subsequently sought care at a pediatric referral for neuromuscular diseases in Argentina, where she was diagnosed with chronic respiratory failure (CRF), oxygen dependence, and intermittent use of nighttime ventilation. She was admitted for treatment of her CRF and returned to Paraguay without oxygen dependence.

She was referred to our hospital, where she received multidisciplinary care with guidance from international experts in the field, following the guidelines for Respiratory Rehabilitation in Neuromuscular Diseases (NMD) studied and published by Bach JR et al. (Table 1). Upon beginning her follow-up with our team, she was trained to use manual bag-valve ventilation to assist her cough using the air-stacking technique, as she presented with a nonfunctional cough characterized by low peak cough flow (PCF) (PCF < 160 L/min) and high levels of exhaled carbon dioxide (eCO_2, 50 mmHg). once the hypercapnia had normalized, serial tracheostomy occlusion was initiated as tolerated; follow-up was interrupted due to hospitalization for a severe respiratory infection. Upon resuming follow-up, a marked decrease in vital capacity was observed, with a loss of more than 400 ml; in previous evaluations, her vital capacity had been greater than 1000 ml, which is why training in lung re-expansion techniques and cough assistance using a mechanically assisted cough (MAC) device was continued (Table 2).

One year after the start of her follow-up by our department, with improved levels of PCF and VC, normal arterial oxygen saturation (SaO_2), and $EtCO_2$, she was successfully extubated (Figure 1). She continued with nocturnal ventilation via noninvasive ventilatory support (NIV) using a binivel pressure device and daily use of the MAC, achieving maximum peak cough flow values of 200 to 250 L/min, with a vital capacity (VC) of 950 ml and normal $EtCO_2$ and SaO_2 values. Following decannulation, the use of the MAC continued, in addition to air stacking, resulting in an improvement in her VC while maintaining normal $EtCO_2$ and SaO_2 values (Table 2).

Table 1. Respiratory rehabilitation guidelines applied

Respiratory Rehabilitation Guidelines for Neuromuscular Disorders (NMD)
1. Regular monitoring of: VC, PCF, $EtCO_2$, and SaO_2.
2. Implement manual bag air stacking technique when:
• VC < 50% predicted or ≤ 2000 mL in adults.
• PCF < 270 L/min.
3. Initiate Mechanically Assisted Cough (MAC) when PCF ≤ 160 L/min.
4. If SaO_2 < 95%: Apply air stacking and MAC until SaO_2 ≥ 95% is restored. If these techniques fail to normalize SaO_2 ≥ 95%, evaluate for hypoventilation, pulmonary disease, pneumonia, or atelectasis.
5. If intubation is required due to severe pulmonary disease, administer MAC hourly until SaO_2 ≥ 95% is maintained without supplemental oxygen and CO_2 levels are normal.
6. In the presence of symptomatic chronic alveolar hypoventilation:
• Prescribe nocturnal NIV.
• Gradually transition to daytime NIV as required

Table 2. Summary of parameters evaluated during the patient's treatment and follow-up

Parameters	1st Eval.	Follow-up	Pre-Extub.	Post-Extub.	At 1 Year
SaO_2	97%	97%	97%	95%	97%
$EtCO_2$	52 mmHg	40 mmHg	39 mmHg	42 mmHg	42 mmHg
VC seated	1009 mL	1100 mL	960 mL	740 mL	900 mL
VC supine	950 mL	950 mL	600 mL	650 mL	730 mL
PCF	N/T	125 L/min	250 L/min	268 L/min	270 L/min

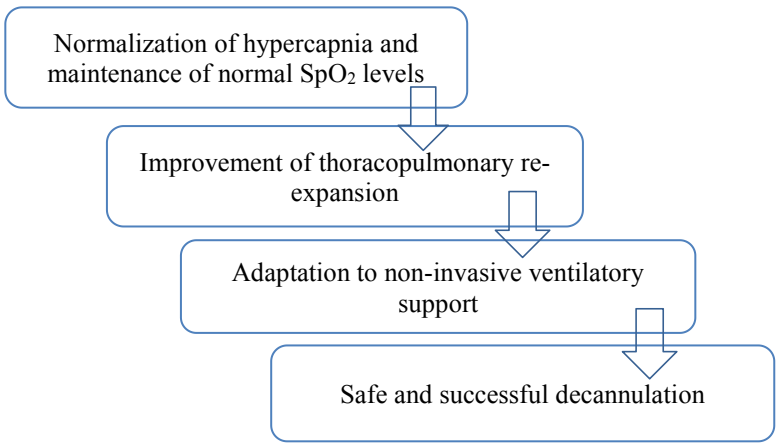

Figure 1. Schematic representation of protocols for successful decanulation in patients with NMD

3 Discussion

The patient-centered respiratory therapy approach focused on the respiratory system, aiming to maintain thoracopulmonary compliance, ensure adequate pulmonary ventilation, optimize cough flow to maintain good airway patency [6], provide training with a mechanical cough assist device to promote safe decannulation [7,8], and provide training with NIV interfaces [9].

A comprehensive evaluation specifically designed for patients with neuromuscular diseases—particularly those with respiratory pump failure—can reduce and even prevent acute respiratory failure (ARF) and tracheotomy [10,11].

Unfortunately, any patient requiring respiratory services—regardless of diagnosis—and is admitted to the intensive care unit will require ventilatory support and a tracheotomy; however, if the patient has a neuromuscular disorder, noninvasive procedures may be chosen [12], thereby optimizing the patient's quality of life as well as reducing hospital costs.

Patients who have been intubated may be extubated and even decannulated with appropriate treatments based on the type of NMD they have [7,8,13].

Respiratory care therapies should not be delayed. To achieve satisfactory ventilatory function, noninvasive ventilatory support (NIV), supplemented and enhanced by a protocol of manual and mechanical assisted coughing, as the preferred option before initiating prolonged mechanical ventilation via tracheostomy [14,15].

Safe and systematic care plays an important role in the patient's recovery. A patient's recovery depends on an entire multidisciplinary healthcare team, with respiratory therapy being of enormous clinical importance, particularly in conditions that compromise respiratory function [16].

The final outcome in this clinical case was decannulation, resulting in an improvement in quality of life. However, as was also the case with this patient, it is very common in the natural history of neuromuscular diseases—even in patients who, despite functional deterioration, are still able to walk—for impairment of the inspiratory respiratory muscles to result in a later need for noninvasive ventilatory support due to respiratory pump failure with progressive symptomatic hypercapnia. However, if standardized protocols for evaluation, follow-up, and intervention are not followed, children, adolescents, and adults with neuromuscular diseases undergo unnecessary tracheostomies when they present with acute upper respiratory infections, which—in the absence of an effective cough due to primary impairment of the expiratory muscles—progress to acute ventilatory failure and subsequently to failure of extubation when an attempt is made to transition from mechanical ventilation to spontaneous breathing without any support [17,18].

These protocols have been synthesized at the Latin American level in publications by the Ibero-American Group for Respiratory Care in Neuromuscular Diseases [3,5,6], whose work has paved the way for the establishment of multidisciplinary centers that integrate with others worldwide (www.breathenvs.com), offering guidance, outreach, and training based on guidelines grounded in the scientific studies and publications of Dr. John Bach (Rutgers University, Newart, New Jersey, USA) [4,7,10-12].

The value of this multidisciplinary network has made it possible to successfully reverse the tracheostomy of a patient with advanced hypercapnic chronic respiratory failure due to congenital myopathy and to transition her from dependence on prolonged mechanical ventilation to the need for noninvasive ventilatory support and assisted coughing protocols , which improve conditions for integration into family, academic, social, and eventually occupational life, with opportunities equivalent to those of other people without physical disabilities. Thus, the objective of this network of centers is to broaden the conceptual framework of treating professionals so they understand that, essentially, no patient with neuromuscular diseases—regardless of age or disease severity, with the exception of those with amyotrophic lateral sclerosis and others with severe bulbar involvement of the upper motor neuron—needs a tracheostomy to survive. To understand that many patients in these clinical conditions and in critical care can be extubated and managed with noninvasive ventilation and assisted coughing without requiring a tracheostomy. To recognize that many tracheostomized patients can be decannulated and managed with noninvasive respiratory care. To recognize that the natural setting for these patients should be their home, with autonomy in decision-making. To disseminate practical tools for the assessment and implementation of

noninvasive management of chronic ventilatory failure. To recognize the differences between noninvasive ventilation and noninvasive ventilatory support and to define the elements of diagnostic support and follow-up.

Conflicts of Interest

The author declares no conflicts of interest regarding the publication of this paper.

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