Acta Scientific MEDICAL SCIENCES (ISSN: 2582-0931)

Volume 7 Issue 8 August 2023

Case Study

Spinal Muscular Atrophy (SMA) Type 1: Case Study of Decannulation

John R Bach[1]*, Louis Saporito[2] and Michael Schepis[3]

[1]Professor of Physical Medicine and Rehabilitation, Professor of Neurology, Rutgers University New Jersey Medical School, USA

[2]Instructor of Physical Medicine and Rehabilitation, Rutgers University New Jersey Medical School, USA

[3]Department of Physical Medicine and Rehabilitation, Rutgers University New Jersey Medical School, USA

***Corresponding Author:** John R Bach, Professor of Physical Medicine and Rehabilitation, Professor of Neurology, Rutgers University New Jersey Medical School, USA.

DOI: 10.31080/ASMS.2023.07.1642

Received: June 20, 2023

Published: July 24, 2023

© All rights are reserved by **John R Bach., *et al.***

Abstract

Three patients with spinal muscular atrophy, who have survived 26 to 30 years by noninvasive respiratory management rather than with tracheostomy tubes, are presented. They demonstrate that tracheostomy tubes are not always necessary for long survival for these patients, even when having 0 ml of vital capacity for decades, that nasogastric tubes do not necessarily prevent effective noninvasive nasal ventilatory support, that patients can be extubated and decannulated of tracheostomy tubes even when having 0 ml of vital capacity, and that noninvasive management is preferred by patients and care providers over the use of airway tubes for ventilatory support.

Keywords: Spinal Muscular Atrophy Type 1; Noninvasive Ventilation; Noninvasive Ventilatory Support; Mechanical Insufflation Exsufflation; Prognosis

Introduction

In 1995 the parents of two infants with spinal muscular atrophy type 1 (SMA1) refused tracheostomy tubes for the infants despite them being intubated and unweanable from continuous invasive ventilatory support. They were told that mortality was certain without a tracheostomy tube because with no skeletal or bulbar muscle function, other than for eye movements, they would not be able to protect their airways and dependence on continuous noninvasive positive pressure ventilatory support (CNVS) had never been described for infants. As a result of refusal of tracheotomy, they and 17 other patients with SMA type 1 are now nasal CVNS dependent and 18 to 30 years old from as young as 4 months of age. None have tracheostomy tubes despite having 0 to 40 ml of vital capacity (VC) and only residual eye movements. However, one of the two patients who first depended on CNVS required emergency tracheotomy at 27 years of age and is presented here.

Case Study

A 28 year old with SMA1 was born April 10, 1995 and diagnosed at 7 weeks of age. He was hypotonic and had paradoxical breathing since birth. A nasogastric (NG) tube was placed at 4 months of age and is still being used over 27 years later despite his dependence

on nasal CNVS for 27 years. He was intubated for acute on chronic respiratory failure at 8 months of age and unweanable from ventilatory support. However, his patients refused tracheotomy and gastrotomy but he was successfully extubated to nasal CNVS on pressure assist control mode, 20 cm H2O inspiratory pressure and no expiratory pressure, entirely against medical advice, including mine. Besides being unlikely to protect his airways, it was mistakenly felt that the NG tube would interfere with nasal NVS. It was also thought that the nasal ventilation would excessively leak out of his mouth.

He weaned to sleep only NVS for 2 months, but by 11 months of age, he no longer had any ventilator free breathing ability (VFBA) and was again CNVS dependent. He was intubated again at 8 years of age for a pneumonia. Mechanical insufflation exsufflation (MIE) was used via the translaryngeal tube (TL) every 2 hours until the ambient air oxyhemoglobin saturation (O2sat) normalized and remained 95% or greater. He was then again extubated to nasal CNVS and MIE and again returned home.

With no functional skeletal or bulbar musculature other than eye movements he has always been averbal. His maximum lifetime VC was 150 ml at age 4 but has been 0 ml since August 2009. Intercurrent respiratory tract infections were successfully treated at home in ambient air by using MIE at 60 cm H2O, via oronasal interface, to clear the airways and reverse all O2 desaturations below 95%.

He had another apparent aspiration pneumonia in August 2021. He was intubated for 6 days and prepared for extubation by using MIE via the tube as described [1,2]. Although again meeting our criteria for successful extubation to CNVS and MIE, which include having normal O2sat in ambient air, upon extubation to CNVS his upper airway closed over a 15 minute period and he underwent emergency tracheotomy to his and his parents' extreme dismay. It was later explained to him that if he preferred continuous tracheostomy mechanical ventilation (CTMV), the tube could remain, or we could remove it for him to return to nasal CNVS [3]. In February of 2022, after 6 months of CTMV, he requested decannulation and was decannulated back to CNVS and MIE in his home with his mother and an experienced nurse present.

With the tube out his MIE exsufflation flows (MIE-EF) exceeded 200 L/m, indicating patency of his upper airway for effective Cough-Assist™ (Phillips Respironics, Murrysville, PA) use to expel airway secretions and maintain normal O2sat. He was successfully decannulated at home. Had his upper airway closed again, the tube could have easily been reinserted for a day or two. He wanted to be decannulated in his home and noted, "because if I needed to do cough assisting (MIE) in my bed, it would be available to me. I had (excessive) airway mucus for approximately 6 weeks (due to the tube). My parents and nurses could cough me with the CoughAssist™ around the clock. It was a successful trach reversal so it's hard to say that I would do it any differently. Having the procedure done in my house was a lot more comfortable than the Dr. office and of course it was safer for me as well".

He noted that he, and his care providers, all prefer nasal CNVS for his sleep, comfort, convenience, and overall, but he preferred CTMV for appearance and security. Since he receives all nutrition via NG since 4 months of age, and has always been averbal, speech and swallowing comparisons were not applicable. He noted that his opinions had not changed since the decannulation. He notes, "having a tracheostomy was painful and scary because I was used to noninvasive ventilation for 26 years. The trach tube was irritating and caused some bleeding. I had increased secretions as well. While I was trached, I used to get very high heart rates and when I first got in my wheelchair, I was so uncomfortable. I had a tracheostomy for a little more than 6 months. I told my parents and Dr. Bach that I wanted to reverse the tracheostomy back to noninvasive ventilation. Noninvasive ventilation is more natural for me. Breathing through my nose was what I was used to doing for so long".

His younger brother, born September 22, 1997, has been more severe. Also, never able to make a vowel or consonant sound, with no skeletal or bulbar muscle function other than for eye but not eyelid movements, he has been nasal CNVS dependent and with an NG tube since 4 months of age and for over 25 years. His CNVS dependence occurred without any hospitalization or episode of respiratory failure at 4 months of age. He had pneumonia at 12 years of age, was intubated, then extubated back to CNVS and MIE. His maximum lifetime VC was 40 ml at 4 years of age.

The other patient with SMA1 for whom tracheotomy was urged at 11 months of age, but refused by his parents, and he is now 30 years old without one. He was born September 28, 1993

and hospitalized and intubated 10 times before 4½ years of age. He was transferred to our units and extubated back to CNVS and MIE 7 times. At 16½ years of age he developed cold, pneumonia, respiratory failure, and was intubated for the eleventh time for 24 hours so that he and his parents could sleep after using MIE on him to clear secretions night and day for 2 weeks. The MIE was often used at 15 to 30 minute intervals during the 2 weeks. Extubated back to CNVS the next day, he again returned home with normal ambient air O2sat and has not been hospitalized again for 13 years.

Two treatment paradigms

Patients with neuromuscular disorders who develop respiratory distress are conventionally treated by supplemental oxygen (O2), bi-level positive airway pressure (PAP) at less than full NVS settings, then with soaring PaCO2 due to the O2 administration [4], they become obtunded and are intubated. When not passing spontaneous weaning trials and ventilator weaning parameters, they are urged to undergo tracheotomy for CTMV or a palliative care death. Over 50% using CTMV, however, die because of the tubes and very few survive to age 20 [5].

A second, noninvasive management paradigm consists of placing symptomatic patients on NVS settings of about 18-24 cm H2O inspiratory pressures (PIPs) without expiratory PAP for sleep. They spontaneously extend use into and, eventually, throughout daytime hours for CNVS as their inspiratory capacities diminish over time. They use CNVS and MIE to clear the airways, along with oximetry feedback to maintain or return O2sat levels to 95% or greater, to avoid intubations during illnesses and when intubated are extubated back to CNVS and MIE. Most such patients become CNVS dependent without hospitalization or episodes of acute respiratory failure. When intubation becomes necessary, MIE is used via the translaryngeal tubes at least every 2 hours until our extubation criteria are satisfied and then post-extubation to maintain normal O2sat. They then return home without tracheostomy tubes [6]. Our center's 18 SMA 1 patients 18 to 30 years of age have a hospitalization rate of 0.02 since after the 10th birthday.

Invasive vs. noninvasive respiratory management

A 1993 publication concerned ventilation interface preferences of 168 patients with ventilatory pump failure (VPF) with at least 1 month experience using both CNVS and CTMV for 17.1 ± 6.5 hours per day and for 22.7 ± 13.1 years in all. The former was preferred for swallowing, sleep, speech, appearance, comfort, convenience, and security by margins of 7 to 9.5 to one and unanimously overall [2]. Since the above case had 26 years of experience using CNVS, then 6 months using CTMV, and another 1½ years back on CNVS and had the option of continuing to use either, he was asked for his preferences. He noted that preferred CNVS overall, but CTMV for appearance. This would likely not have been the case if he were able to use mouthpiece NVS or an intermittent abdominal pressure ventilator which has not yet been available to him [7]. He noted that he preferred tracheotomy for security. This was due to inability to perform glossopharyngeal breathing for VFBA in the event of ventilator failure or disconnection which 70% of CNVS users with functional bulbar innervated musculature can do [8]. There can be no more relevant perspective on the desirability of NVS over TMV for quality of life than by Case 1.

Concerning decannulation of patients with no VFBA and 0 ml of VC to nasal CNVS, we do not recommend that this be done in their homes, or for children under age 12. The point is, however, that when prepared for extubation by using MIE via the tube until room air O2sat is normal, as per criteria [3,9,10], extubation or decannulation to CNVS and MIE can be routine whether patients can breathe unaided or not [3,9,10]. Many of our patients with little to no VC have been decannulated in the outpatient clinic rather than in a hospital after practicing nasal NVS at home using a capped cuffless fenestrated tracheostomy tube [10].

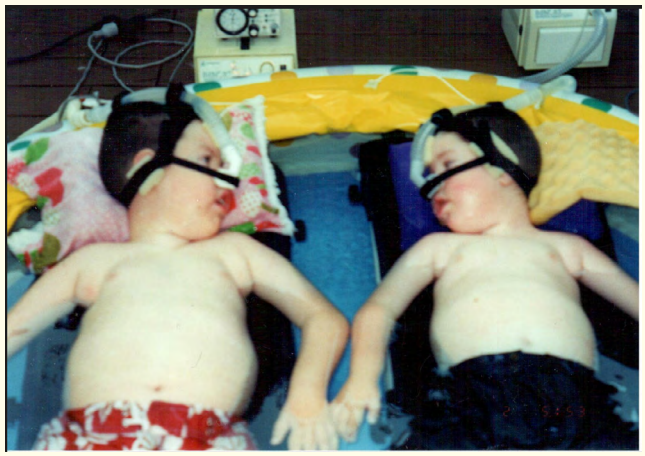

Figure 1

Figure 2

Figure 4

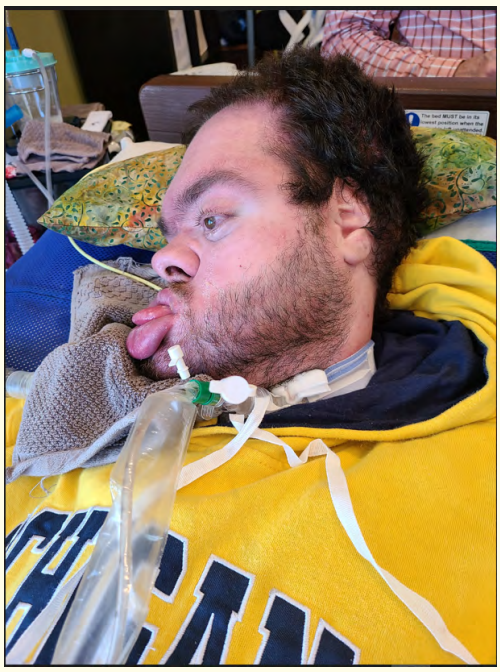

Figure 3

Figure 5

Conclusion

In conclusion, 1) NG tubes do not necessarily prevent effective nasal CNVS for patients, even those with 0 ml of VC. 2) Patients with no VFBA and 0 ml of VC can be routinely extubated and decannulated of tracheostomy tubes, although we do not recommend that this routinely be done in their homes. 3) Patients with SMA1 can survive for decades, even without the new upstream medications that can make them stronger when available to them. 4) Noninvasive management of VPF is preferred by patients and their care providers over tracheostomy ventilation [11,12].

Bibliography

1. Bach JR. "Point: Is Non-invasive ventilation always the most appropriate manner of long-term ventilation for infants with spinal muscular atrophy type 1? Yes, almost always?" *Chest* 151.5 (2016): 962-965.

2. Bach JR. "A comparison of long-term ventilatory support alternatives from the perspective of the patient and care giver". *Chest* 104.6 (1993): 1702-1706.

3. Bach JR., *et al*. "Extubation of unweanable patients with neuromuscular weakness: a new management paradigm". *Chest* 137.5 (2010): 1033-1039.

4. Chiou M., *et al*. "Quantitation of Oxygen induced hypercapnia in respiratory pump failure". *Revista Portuguesa de Pneumologia, Portuguese Journal of Pulmonology* 22.5 (2016): 262-265.

5. Carter RE., *et al*. "Comparative study of electrophrenic nerve stimulation and mechanical ventilatory support in traumatic spinal cord injury". *Paraplegia* 25 (1987): 86-91.

6. Bach JR., *et al*. "Decannulation of patients with severe respiratory muscle insufficiency: efficacy of mechanical insufflation-exsufflation". *Journal of Rehabilitation Medicine* 46 (2014): 1037-1041.

7. Bach JR and Alba AS. "Intermittent abdominal pressure ventilator in a regimen of noninvasive ventilatory support". *Chest* 99.3 (1991): 630-636.

8. Bach JR. "New approaches in the rehabilitation of the traumatic high level quadriplegic". *American Journal of Physical Medicine and Rehabilitation* 70.1 (1991): 13-20.

9. Bach JR., *et al*. "Efficacy of mechanical insufflation-exsufflation in extubating unweanable subjects with restrictive pulmonary disorders". *Respiratory Care* 60.4 (2015): 477-483.

10. Bach JR., *et al*. "Decanulation of patients with severe respiratory muscle insufficiency: efficacy of mechanical insufflation-exsufflation". *Journal of Rehabilitation Medicine* 46 (2014): 1037-1041.

11. Bach JR. "A comparison of long-term ventilatory support alternatives from the perspective of the patient and care giver". *Chest* 104.6 (1993): 1702-1706.

12. Bach JR., *et al*. "Spinal muscular atrophy type 1 quality of life". *American Journal of Physical Medicine and Rehabilitation* 82.2 (2003): 137-142.