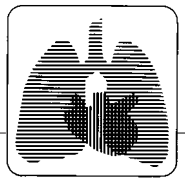

Spinal Muscular Atrophy Type 1*

A Noninvasive Respiratory Management Approach

John R. Bach, MD, FCCP; Vis Niranjan, MD; and Brian Weaver, BS, RRT

Study objective: **To determine whether spinal muscular atrophy (SMA) type 1 can be managed without tracheostomy and to compare extubation outcomes using a respiratory muscle aid protocol vs conventional management.**
Design: **A retrospective cohort study.**
Methods: **Eleven SMA type 1 children were studied during episodes of respiratory failure. Nine children required multiple intubations. Along with standard treatments, these children received manually and mechanically assisted coughing to reverse airway mucus-associated decreases in oxyhemoglobin saturation. Extubation was not attempted until, most importantly, there was no oxygen requirement to maintain oxyhemoglobin saturation greater than 94%. After extubation, all patients received nasal ventilation with positive end-expiratory pressure. Successful extubation was defined by no need to reintubate during the current hospitalization.**
Results: **Two children have survived for 37 and 66 months and have never been intubated despite requiring 24-h nasal ventilation since 5 and 7 months of age, respectively. One other child underwent tracheostomy for persistent left lung collapse and inadequate home care, another for need for frequent readmission and intubation, and one child was lost to follow-up 3 months after successful extubation. The other six children have been managed at home for 15 to 59 (mean 30.4) months using nocturnal nasal ventilation after an episode of respiratory failure. The nine children were successfully extubated by our protocol 23 of 28 times. The same children managed conventionally were successfully extubated 2 of 20 times when not using this protocol ($p < 0.001$ by the two-tailed Fisher's Exact *t* Test).**
Conclusion: **Although intercurrent chest colds may necessitate periods of hospitalization and intubation, tracheostomy can be avoided throughout early childhood for some children with SMA type 1.** ***(CHEST 2000; 117:1100–1105)***

Key words: bilevel positive airway pressure; mechanical ventilation; noninvasive ventilation; pulmonary complications; respiratory failure; spinal muscular atrophy; survival

Abbreviations: EPAP = expiratory positive airway pressure; IPAP = inspiratory positive airway pressure; MI-E = mechanical insufflation-exsufflation; Sao_2 = arterial oxyhemoglobin saturation; SMA = spinal muscular atrophy

Autosomal recessive spinal muscular atrophy (SMA) is the most common inherited neuromuscular disease of the hypotonic newborn and, along with Duchenne muscular dystrophy, is 1 of the 2 most commonly inherited neuromuscular diseases. It is caused by a chromosome 5 defect. About 1 in 40 people carry the defective gene, and the overall incidence has been reported to be 1 out of 5000.[1] It has been categorized into 4 types according to severity. The SMA type 1 infant never attains the ability to sit independently. Less than 20% of these children survive 4 years, and then only with indwelling tracheostomy tubes.[2] Virtually all die from respiratory complications. SMA type 2 children can temporarily sit independently but can never walk, and they too usually have periods of respiratory failure during early childhood. Other SMA types have milder courses.

*From the Department of Physical Medicine and Rehabilitation (Dr. Bach), the Department of Pediatrics (Dr. Niranjan), University of Medicine and Dentistry of New Jersey-New Jersey Medical School; and University Hospital (Mr. Weaver), Newark, NJ.
This work was performed at the University Hospital of the University of Medicine and Dentistry of New Jersey-New Jersey Medical School in Newark, NJ.
Manuscript received February 8, 1999; revision accepted October 26, 1999.
Correspondence to: John R. Bach, MD, FCCP, Department of Physical Medicine and Rehabilitation, University Hospital B-403, 150 Bergen Street, Newark, NJ 07103; e-mail: bachjr@umdnj.edu