Breathing Basics and Care Choices for SMA Type I

2015 Annual Cure SMA Conference
Westin Hotel – Kansas City, MO
June 18-21, 2015
PRESENTERS

- **Mary Schroth MD**  
  University of Wisconsin School of Medicine and Public Health  
  American Family Children’s Hospital  
  Madison, Wisconsin

- **Richard Shell MD**  
  The Ohio State University  
  Nationwide Children’s Hospital  
  Columbus Ohio

- **Richard Kravitz MD**  
  Duke University School of Medicine  
  Duke University Medical Center  
  Durham, North Carolina
Encompasses:

• Diagnosis
• Respiratory Care
• GI and Nutrition
• Orthopedic Concerns
• Palliative Care
SMA MYTHS

• You have no hope
• You have no choices
• There is nothing you can do
• Your child is going to die
• You have no support
• A tracheostomy is a sign of failure
SMA FACTS

• Treatment options are available (though they may be labor intensive and are more supportive, not curative)

• Children with SMA of all types are doing better than ever before

• There is currently no cure, but research is underway and therapies might yet be on the horizon

• A tracheostomy represents just another a treatment modality
Results of Respiratory Muscle Weakness in SMA

1. Difficulty coughing
2. Small shallow breaths during sleep: hypoventilation
3. Chest wall and lung underdevelopment
4. Recurrent infections that contribute to muscle weakness.

Other Complications of Neuromuscular Weakness

- Scoliosis
  - Alters chest wall shape
- Gastroesophageal reflux and aspiration
- Constipation
Chest Wall Changes

Normal

SMA I
Normal breathing

Respiratory and bulbar muscle weakness

REM related sleep disordered breathing

Ineffective cough reduced peak cough flows

NREM and REM sleep disordered breathing

Swallow dysfunction

Chest infections

Daytime ventilatory failure

Physical examination

Pulmonary function, peak cough flow, respiratory muscle strength

Chest xray, Sleep study

Swallow function evaluation

Airway clearance with cough assistance

Nocturnal non-invasive ventilation

Nocturnal or continuous non-invasive ventilation

FRC Relative to Position for Diaphragm Dependent Breathers

From Nunn’s Applied Respiratory Physiology, 2000
IMPACT OF MUSCLE WEAKNESS

- Scoliosis
curvature of the spine

- Restrictive lung disease
decreased lung volumes

- Weakened cough

- Increased risk of aspiration
  and gastroesophageal reflux

- Atelectasis
  mucus plugging

- Pneumonia

- Hypoventilation
  inadequate breathing while
  asleep/awake

- Obstructive sleep apnea
CAUSES OF RESPIRATORY PROBLEMS

- Inadequate cough
  - Retained secretions
  - Mucus plugging
  - Atelectasis
  - Hypoxia (low $O_2$)
  - Respiratory distress
  - Respiratory failure

- Infection

- Inadequate ventilation
  - Hypoxia (low $O_2$)
  - Hypoventilation
  - Respiratory distress
  - Respiratory failure

- Cough and Ventilation Worsen With Respiratory Illnesses!
METHODS TO ASSESS RESPIRATORY MUSCLE FUNCTION

• Pulmonary function testing
  Spirometry
  Lung volumes

• O₂/CO₂ analysis
  Blood gas (arterial, venous, capillary)
  Pulse oximetry (SaO₂)
  Capnography (EtCO₂)

• Maximal respiratory pressures
  Maximal Expiratory Pressure
  Maximal Inspiratory Pressure

• Cough Peak Flow

• Chest x-ray

• Overnight oximetry/capnography

• Polysomnogram (sleep study)
FACTORS AFFECTING SECRETIONS

- Quantity of secretions
  Normal versus increased amount
  Ability of cough to remove a usual amount of secretions

- Quality of secretions
  Normal (watery) versus sticky
  Infection (viral versus bacterial)
INTERVENTIONS TO AID AIRWAY CLEARANCE

• Secretion mobilization (non-pharmacologic)
  – Manual chest physiotherapy
  – Intrapulmonary percussive ventilation (IPV)
  – High frequency chest wall oscillation (*Vest therapy*)
  – (bronchoscopy)

• Mucociliary clearance (pharmacologic)
  – Albuterol; hypertonic saline 3 or 7%; Pulmozyme

• Cough augmentation
SECRETION MOBILIZATION
Non-pharmacologic

• Helpful in SMA
  Manual chest physiotherapy
  High frequency chest wall oscillation
    (Vest therapy)
  Intrapulmonary percussive ventilation (IPV)
    (Bronchoscopy)

• Less helpful in SMA
  Positive Expiratory Pressure
    Acapella
    Flutter
Postural Drainage
COUGH MECHANISM

3 Phases of a cough

1. Inspiratory phase
2. Closure of vocal cords/contraction of expiratory muscles
3. Opening of the vocal cords
FACTORS IMPAIRING A COUGH

- **Inadequate inhale** (need to get to 85-90% of Lung Capacity)
  - Inherent muscle weakness
  - Muscle fatigue
  - Scoliosis
  - Chest wall stiffness

- **Incomplete closure of throat and vocal cords**
  - Not usually a SMA problem unless a tracheostomy is needed

- **Inadequate exhale** (need cough flow rate > 6L/min)
  - Same problems with inspiration
  - **PLUS**
  - Diminished Inspiratory Capacity → Mechanical disadvantage
    - Muscles not at point of maximal capacity for active contraction
    - Lung not at point of maximal (passive) elastic recoil
    - Airway diameter not fully maximized
MECHANICAL IN-EXSUFLATOR
(THE COUGH ASSIST)

• Well tolerated:
  No gastroesophageal reflux or aspiration
  No pneumothorax (collapsed lung)

• Main complaints: “I just don’t like it!”
  Interface discomfort
  Pressures uncomfortable
  Chest/abdominal discomfort
  No perceived benefit

• Most effective when used regularly
  (versus only when ill)
  Takes some practice, but few complaints
  Once used to it, most kids will start to ask for it when ill!
Machine for Coughing
Chest radiograph of a 22-month-old with SMA type I.
THE COUGH ASSIST

• Settings for use:
  – Inspiratory:
    • visually good lung expansion (up to +40 cm H2O)
    • Over 1-2 seconds
  – Expiratory:
    • start at -25 cm H₂O (up to -40 cm H₂O)
    • Over 1-2 seconds
  – Pause: 1-2 seconds between cycles

• Number of cycles:
  – 3-5 breaths/cycle
  – 3 to 5 cycles per treatment (as tolerated)

• Frequency of use:
  – **When well:** once daily to twice a day (as needed)
  – When ill: as often as necessary
THE COUGH ASSIST

• Have suction available to help patient expectorate sputum (try to avoid deep suctioning)

• Various interfaces acceptable (full face mask; mouth piece; endotracheal tube; tracheostomy tube)

• Observe for transient $O_2$ desaturations with use (suggests mucus mobilization)
OTHER FACTORS AFFECTING AIRWAY CLEARANCE

- **Nutrition**
  - Obesity
  - Malnutrition

- **Sleep**
  - Hypoventilation
  - Obstructive sleep apnea

- **Aspiration**
  - Video Swallowing Study

- **Gastroesophageal reflux**
ASPIRATION-RELATED FACTORS

- Aspiration
  - Impaired swallowing
  - Impaired protective reflexes
  - Gastroesophageal reflux

- Gastroesophageal reflux
  - Decreased lower esophageal muscle tone
  - Scoliosis can make the esophagus pass through the diaphragm at a bad angle, leading to reflux
  - Poor stomach emptying
ASPIRATION-RELATED FACTORS TREATMENTS

- Modified feeding plan
  - Speech therapy input
  - Thickened feeds
  - Tube feeding

- Drying agents
  - Glycopyrrolate (Robinul)
  - Botulism toxin injections

- Thickened feeds
  (if oral feeds are tolerated)

- Acid Blockers
  - H₂ blockers
  - Proton pump inhibitors

- Surgery
  - Gastrostomy tube +/- Nissen fundoplication
  - Parotid duct ligation
RESPIRATORY SYMPTOM PROGRESSION

- Normal (asymptomatic)

- Weakened cough
  - Usually adequate when well
  - Problems develop with infections
  - Can lead to (recurrent) pneumonia/bronchitis

- Nighttime symptoms

- Daytime symptoms
RESPIRATORY INTERVENTION PROGRESSION

- Normal (asymptomatic)
  - No treatment needed

- Weakened cough
  - Assist in airway clearance (required with viral illnesses)
    - Assisted cough
    - Chest physiotherapy
  - May require ventilation (if low $O_2$ levels or in respiratory distress)

- Nighttime symptoms
  - BiPAP or volume ventilator when asleep

- Daytime symptoms
  - Continuous ventilation
  - Non-invasive ventilation versus tracheostomy (in select patients)
Pulse Oximetry

- Normal is $\geq 95\%$ or greater
- $< 95\%$ suggests:
  - Mucus plugging (needs more airway clearance)
  - Shallow breathing (awake and/or asleep)
Breathing Support Approaches

- Non-invasive ventilation
  BiPAP or Ventilator
  Interface: Mask over the nose

- Invasive ventilation
  Ventilator
  Interface: Tracheostomy

- Comfort Care (as part of Palliative Care)
GOALS OF VENTILATORY SUPPORT

• Improve oxygen and carbon dioxide levels
  Ventilation and Oxygenation

• Muscle rest

• Treatment of pectus excavatum

• Compensate for worsening strength when having a respiratory illness
Non-Invasive Positive Pressure Ventilation Devices

- Bi-level positive airway pressure (BiPAP)
- Home ventilator
BREATHING ASSIST (Interfaces)

- Non-invasive Ventilation
- Invasive Ventilation
POSTITIVE PRESSURE VENTILATION: Advantages of positive pressure

- Advantages
  - Non-invasive
  - Reversible
  - Safe
  - Effective
  - Preserves (natural) airway protection mechanisms
  - Decreased risk of infection
  - Comfort
  - Preserves speech
  - Less swallowing issues

- Disadvantages
  - Lack of access to airway for deep suctioning
  - Aerophagia
  - (worsened reflux)
  - Facial injury
  - Facial bone remodeling from tight fitting mask
  - Increased oral secretions
  - Requires cooperation of the patient
NIPPV COMPLICATIONS

- Nasal/oral dryness
- Epitaxis
- Nasal congestion
- Sneezing
- Rhinorrhea
- Sinusitis

- Claustrophobia
- Mask irritation
- Nasal abrasions
- Aerophagy
- Mouth leak
- Facial deformities
- (Decreased cardiac output)
INVASIVE RESPIRATORY CARE

- Intubation (in an acute illness)
- Tracheostomy with ventilator support
TRACHEOSTOMY: Indications

• NIPPV no longer effective at treating hypoventilation

• NIPPV not tolerated by the patient

• Excessive oral secretions puts patient at risk for aspiration

• Care for patient with NIPPV beyond the capabilities of the family

• Resources for outpatient management with NIPPV not available in the community

• Failure to extubate a patient who has been intubated (for whatever reason). This assumes proper methods of extubation were employed
TRACHEOSTOMY: Advantages for the Patient

- Allows for assisted ventilation
- Airway is protected from aspiration
- Increased ease in suctioning excessive secretions (if needed)
- No NIPPV mask pressure sores or facial remodeling
- Treats obstructive sleep apnea (if present)
- In select cases, may be a temporizing measure until extubation is possible
TRACHEOSTOMY:
Advantages for Family

• Freedom
  – night out for the parents
  – more mobility
    • getting to doctor’s office/hospital
    • shopping
    • vacations

• Increased support in the home
  – more skilled nursing care
  – more nursing aid hours
  – (make sure you have a good case manager)
TRACHEOSTOMY: Disadvantages

- Unsightly
- Diminished cough
- Risk of infection
- Secretions
- Speech problems
  - trouble understanding child
  - never develop speech
  - loose ability to speak
- Dysphagia
- Bradycardia
- Desaturations
- Loss of airway (if the tube comes out)
- Increased hospitalizations

- Suprastomal damage
  - granulation tissue
  - airway collapse
- Subglottic damage
  - granulation tissue
  - tracheomalacia
  - tracheomegaly
  - tracheal erosions
  - fistula formation
  - subglottic stenosis
- Bleeding
  - suction catheter trauma
  - arterial erosion
TRACHEOSTOMY: Quality of Life Issues

- The family unit
  - limited data (especially with children)
  - the perspective varies:
    - the patient
    - the parents
    - the siblings

- The medical team
  - health care providers ≠ the family
  - *health care providers frequently underestimate the patient’s quality of life!*
KEY POINTS REGARDING A TRACHEOSTOMY

• Elective tracheostomy is better than emergency tracheostomy

• Long-term treatment plan is best determined in times of health. The middle of an acute decompensation (especially one which was predictable) is not the best time to be making these types of decisions
PALLIATIVE CARE

• Recognizes that SMA Type I is a life threatening disease

• Should be part of the care of the disease process

• Goals
  Provide symptom relief: pain, dyspnea, agitation, nausea, anxiety
  Provide psychological, social and spiritual support for patient and family

• Palliative care can be in the hospital, in the home, or both

• Palliative care is not giving up; it is just changing the focus of therapy
HOW THE COMMON COLD MAKES CHILDREN WITH SMA SICK

- Increased secretions
  Worsening of (already impaired) swallowing function
  Increased risk of aspiration

- Weakened cough
  Diminished ability to clear secretions
  Increased risk for mucus plugging, atelectasis, and pneumonia
  Hypoxia

- Further weakening of muscle strength
  Vicious cycle leading to more aspiration and further weakening of an already diminished cough
  Higher risk for inadequate nighttime breathing and hypoxia
HOW TO TREAT THE COMMON COLD

• Treat the increased secretions
  Good hydration
  Suctioning (if necessary)
  Avoid drying agents/anti-histamines (if possible)

• Treat the weakened cough
  Increased use of cough assisting maneuvers (i.e.: the Cough Assist)
  You can do this as often as needed (up to every 10-15 minutes)
  Maintain normal oxygenation (SaO₂ ≥ 95%) **Avoid supplemental O₂**!
  Assisted ventilation may be needed
  Judicious use of antibiotics

• Treat the worsened muscle strength
  The patient may require increased use of NIPPV (both while awake and asleep)
  *Watch out for worsening nocturnal hypoventilation*
ISSUES TO ADDRESS AT FOLLOW UP APPOINTMENTS

- Pulmonary
- Sleep
- GI status
- Well child care
WELL CHILD CARE

• Routine health care

• Immunizations
  Usual immunization
  Influenza vaccination
  Pneumococcal vaccination
  Palivizumab (Synagis)

• Proper nutrition
  Obesity versus malnutrition
EQUIPMENT RECOMMENDED FOR THE HOME

- Mechanical in-exsufflator (The Cough Assist)
- Ambu bag
- Pulse oximeter
- Chest cups or high frequency chest wall oscillation (if needed)
- Suctioning equipment
- Nebulizer (if needed)
- BiPAP or volume ventilator (when needed)
Children with SMA type I are very weak in the first year of life and their prognosis is not clear.

Children with SMA type 1 are at great risk for poor cough and hypoventilation.

Not all interventions fit every child.

You will make the best decisions for your child.
WHEN THE OPTIONS ARE LIMITED

• Families often have the difficult position of choosing the option they believe is best for their child and their family

• Because there are no right or wrong choices, you must trust in yourselves that you will make the right decision
MULTIDISCIPLINARY HEALTH CARE TEAM

- Pulmonologist
- Neurologist
- Cardiologist
- Orthopedist
- Sleep physician
- 1° Care physician
- Nursing
- Nutritionist

- Respiratory therapist
- Physical therapist
- Occupational Therapist
- Speech Therapist
- Social work
- Palliative care
THINGS TO REMEMBER

• Every child is unique
• You are your child’s best advocate
• The decisions you make regarding your child’s care are the correct ones for your family
• You are part of team caring for your child (actually, you are the captain, so the call is yours), but don’t be afraid to ask for input and be open to your health care team’s recommendations
• You are not alone. There are many support groups out there to help you.
• Get involved!
REMEMBER, ANYTHING IS POSSIBLE…
Please complete your conference survey at this link:
https://www.surveymonkey.com/s/2015AnnualSMAConference

Or fill out the paper survey in your conference folder.

All participants who complete the survey will receive a raffle ticket to win an IPad! Winner will be announced on Sunday, June 21st at the Closing General Session.