

Breathing Strong All Day Long

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Objectives

- Understand basics of breathing
- Understand how SMA affects breathing
- Understand the impact a cold can have on breathing
- Learn techniques to help your child's breathing
- Understand Non-Invasive and Invasive ventilation
- Future directions

Consensus Statements

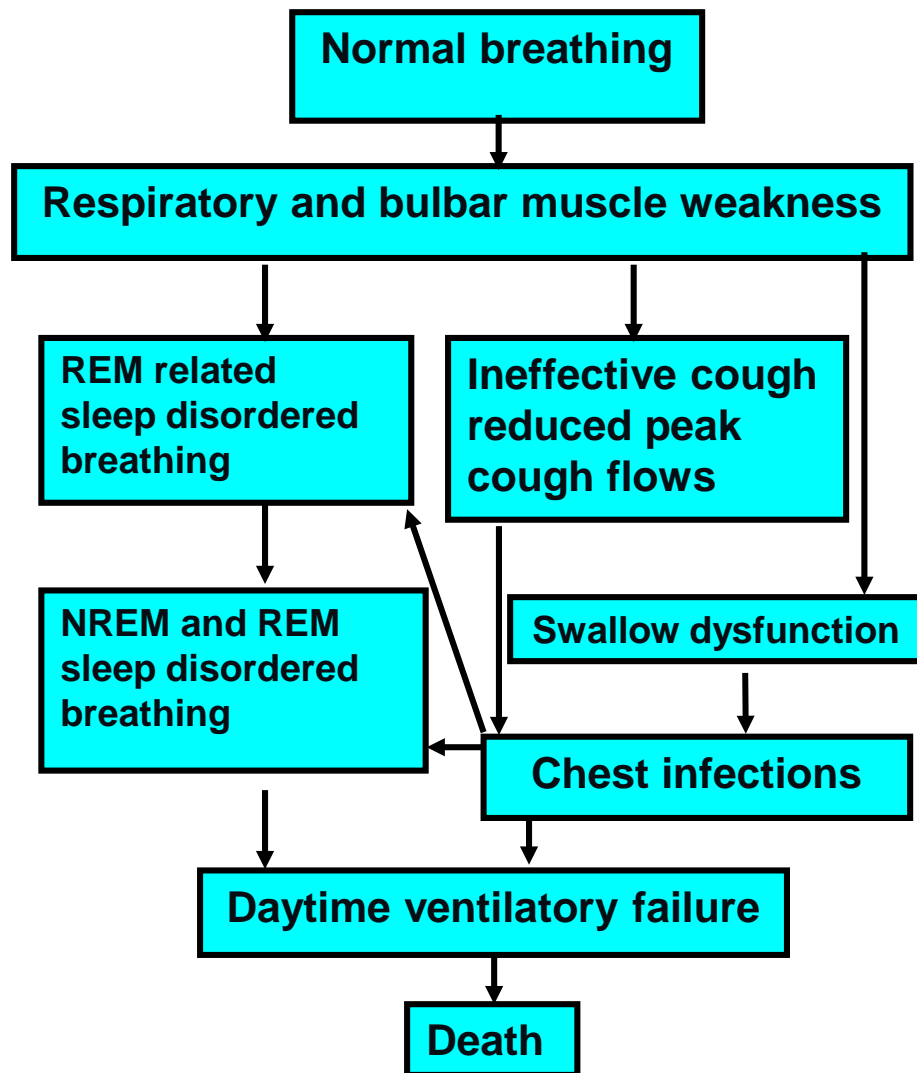
- Consensus Statement in the Standard of Care in Spinal Muscular Atrophy, Wang C et al, J Child Neurol 2007; 22:1027
- 1st Italian SMA Family Association Consensus Meeting: Management and Recommendations for the Respiratory Involvement in Spinal Muscular Atrophy types I-III Rome, Italy 30-21 January 2015, Sansone V.A. et al, Neuromuscular Disorders 25 (2015) 979-989
- Diagnosis and management of spinal muscular atrophy: Part 2: Pulmonary and acute care; medications, supplements and immunizations; other organ systems; and ethics, Finkle R.S., et al Neuromuscular Disorders, Neuromuscular Disorders 28 (2018) 197–207

Results of Respiratory Muscle Weakness in SMA

1. Impaired respiratory “pump” leads to night time and then daytime hypoventilation with increased CO₂ levels
2. Impaired cough and poor clearance of airway secretions
3. Recurrent chest infections exacerbating muscle weakness
4. Swallowing dysfunction and reflux leading to aspiration
5. Upper airway obstruction causing nighttime desaturation
6. Paradoxical breathing leading to chest wall and lung under development
7. Progressive scoliosis with “mechanical” effects on breathing

SMA Pulmonary

Natural History



Natural History

Assessment

Physical examination

Pulmonary function, peak cough flow, respiratory muscle strength

Chest xray, Sleep study

Swallow function evaluation

Intervention

Airway clearance with cough assistance

Nocturnal non-invasive ventilation

Nocturnal or continuous non-invasive ventilation

Evaluation of the Respiratory System

- Lung Function Measurement
 - Vital Capacity (Forced Vital Capacity = FVC)
 - Total Lung Capacity (TLC)
 - Residual Volume (RV)
- Static Pressures
 - Maximal Inspiratory Pressure (cmH₂O)
 - Maximal Expiratory Pressure
 - Sniff Nasal Inspiratory Pressure (SNIP)

Evaluation of the Respiratory System

- Cough Peak Flow or Cough Peak Expiratory Flow
- Capillary Blood gas or End Tidal CO₂
- Pulse oximetry (O₂ sat)
- Sleep studies (PSG)
- Chest Xray

Basics of Breathing

- Diaphragm is main muscle of breathing
 - Inhalation is active (diaphragm contracts)
 - Exhalation is passive (diaphragm relaxes)
- Force exhalation and coughing require the use of accessory muscles
 - Chest wall muscles (intercostal muscles)
 - Abdominal wall muscles
 - Other muscles
- To have an effective cough need all the muscles to work and need to be able to take a big breath

What leads to Abnormal Breathing?

- Breathing spontaneously is a balancing act between the ability of the breathing muscles on one side and the forces of the lung tissue, chest wall and airways on the other side all controlled by the brain's central drive to breath
 - Normal respiratory muscle strength can be overcome by abnormal airways, stiff lungs or stiff chest walls
 - Leads to respiratory failure
 - Weak muscles can be overcome by the forces of normal lung/chest wall/airways
 - Leads to respiratory failure
 - Poor drive to breathe by the brain also causes abnormal breathing
 - Caused by narcotics, general anesthesia or other medications
 - All of the above result in elevated CO₂ and decrease O₂ saturation

Respiratory Consequences of SMA

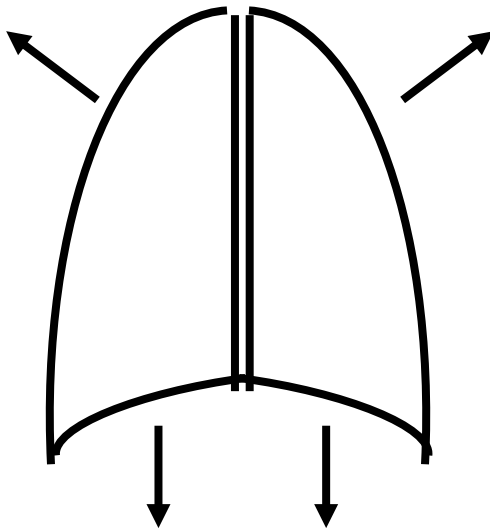
- Weak intercostal muscles and relatively stronger diaphragm
 - Diaphragm is the primary breathing muscle in patients with SMA I and II
- Impaired cough results in poor clearance of lower airway secretions
- Sleep disordered breathing is common
 - Elevated CO₂ is common (hypoventilation)
- Chest wall and lung underdevelopment commonly occurs
- Recurrent infections can worsen muscle weakness and lead to damage to the lung tissue.

Respiratory Consequences of SMA

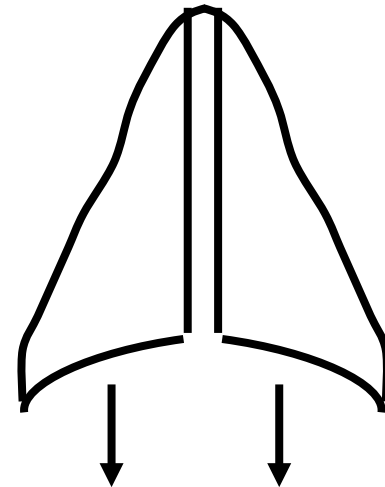
- Weak accessory muscles with spared diaphragm
 - leads to a bell shaped chest with abnormal rib placement and depressed sternum
 - Lung under-development / decreased lung growth
 - Mechanical disadvantage
 - Paradoxical breathing

Chest Wall Changes

Normal



SMA 1 and
some type 2



Respiratory Consequences of SMA

- Impaired cough due to weak expiratory muscle function results in poor clearance of lower airway secretions
 - Causes airway obstruction
 - Decreased lung function
 - Bronchitis or Pneumonia
 - Decreased O₂ saturation

Pulse Oximetry

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



Respiratory Consequences of SMA

- Sleep disordered breathing is common with decreased O2 sats and elevated CO2
- Normal breathing in sleep is a highly regulated process with sensors in the major blood vessels and the brain which signals the body to keep O2 and CO2 within normal limits
 - Balance between respiratory rate, depth of breathing
 - Can see slightly decreased O2 and increased CO2 in normal people during sleep
 - Loss of muscle tone during sleep leads to obstruction, hypoventilation (under breathing)

Respiratory Consequences of SMA

- A Sleep Study (PSG) measures:
 - respiratory rate, HR, CO₂, EEG, O₂ sats, airflow from nose and mouth, eye muscle movement, chest and abdominal wall motion, leg movement
 - Stages of sleep include Non-REM and REM
 - REM sleep (rapid eye movement)
 - Increased intercostal muscle weakness leads to paradoxical breathing
 - Can see paradoxical breathing even with a relatively normal PSG
 - More problems seen in REM sleep

SMA type I/Nonsitters

- Weak intercostal muscles
- Chest wall: very soft and flexible during the first year of life
- Diaphragm: easily fatigued, the primary muscle for breathing
- Other complications:
 - Dysautonomia
 - Dysphagia with aspiration
 - Scoliosis, joint contractures
 - Poor bone quality – increased fracture risk
 - Intermittent gastroparesis

SMA type II/Sitters

- Range of respiratory muscle weakness
 - Weak intercostal muscles
 - Chest wall: rib collapse over time (parasol deformity)
 - Diaphragm: fatigue, primary muscle for breathing
- Other complications:
 - Some develop dysphagia – can occur in teens
 - Scoliosis, joint contractures
 - Poor bone quality – increased fracture risk
 - Chronic pain
 - Obesity as they get older

SMA type III/Walkers

- Generally normal pulmonary function tests
- At risk for:
 - Obstructive sleep apnea
 - Respiratory muscle weakness in adolescence and adulthood
 - Respiratory compromise with anesthesia, narcotic use, illness
- Other complications:
 - Obesity
 - Scoliosis, joint contractures
 - Chronic pain

Summary

- Caretakers need to understand basics of breathing
- Caretakers need to understand respiratory consequences of SMA
- Need to obtain regular evaluation of the respiratory system
 - Things can change over time!!
 - Understand limits of current testing
- Early intervention in respiratory infections, or worsening disease is essential

Thank you very much !!





cure
SMA

Breathing Support

- Why do we do it?
 - What are your goals and can breathing support help you get there?
- When do we use it?
 - How do we know we need it?
- How do we do it?
 - A few different ways

Goals of breathing support

- Longer life
- Muscle rest so strength can be used elsewhere
 - Normal oxygen
 - Normal carbon dioxide
- Quality of life
 - Better sleep = happy person
 - Reduce hospitalizations
- Acute care – support during illness
- Treatment of pectus excavatum (caved in chest)

Chest wall development

6 months



18 months



Courtesy of A. Simonds, Royal Brompton Hospital, UK

Pulmonary function tests

- Forced vital capacity (FVC): How much air can I blow out after taking the deepest breath
- Peak cough flow (PCF): How fast does the air come out when I cough?
- Maximal inspiratory pressure (MIP): How hard can I inhale?
- Maximal expiratory pressure (MEP): How hard can I exhale?
- *Limited by age and strength*

When to start respiratory support

- FVC less than 50% predicted
- MIP less than 60 cm H₂O
- Oxygen less than 95% persistently
- CO₂ over 45 mmHg
- Sleep study showing high CO₂, low O₂, fast breathing, poor sleep structure
- Recurrent respiratory infections requiring hospitalization
- **SIGNS OR SYMPTOMS OF IMPAIRED SLEEP**
 - Restlessness, frequent awakening, sweating, morning headaches, daytime fatigue, “fading” in the afternoon...

When to start respiratory support

- What about a sleep study?
 - Problematic: most labs set up for sleep apnea, not SMA/hypoventilation so diagnostic and titration studies may miss important features specific to weakness
- How to determine settings
 - Sleep study – limited access, not always set up for SMA
 - Good clinical reasoning and experience – not scientific
 - Data downloads from your device can be very helpful
 - Show respiratory rate, breath size, effort, duration of use

How to provide breathing support - positive pressure devices

Bi-level positive airway pressure devices – Non- invasive only



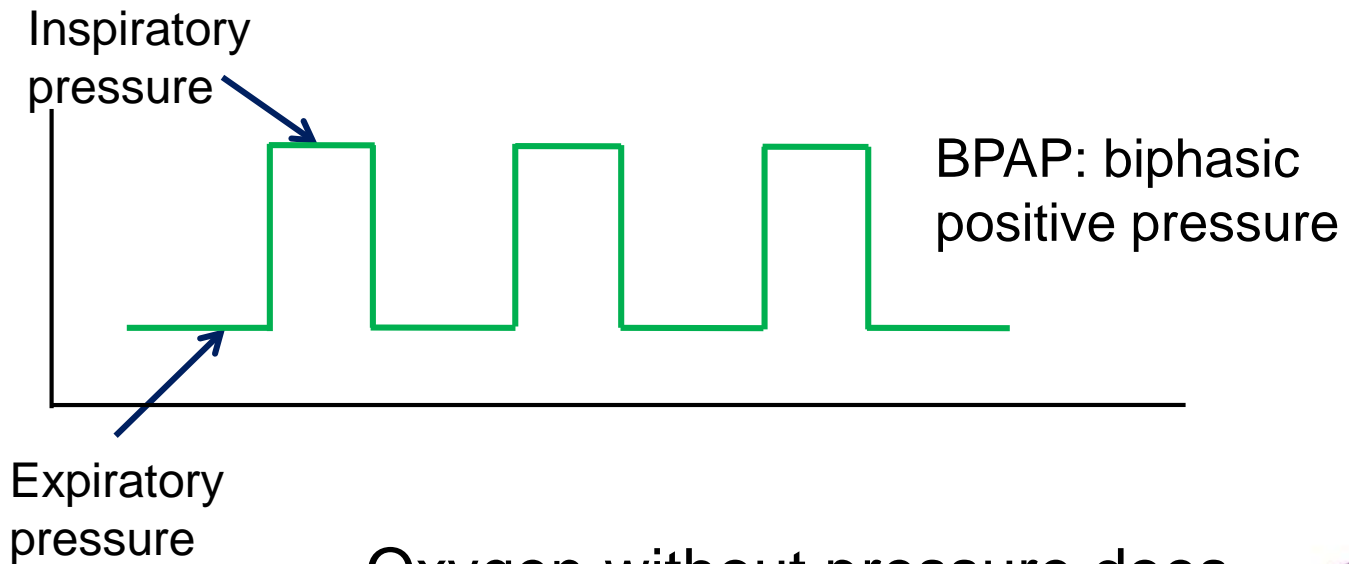
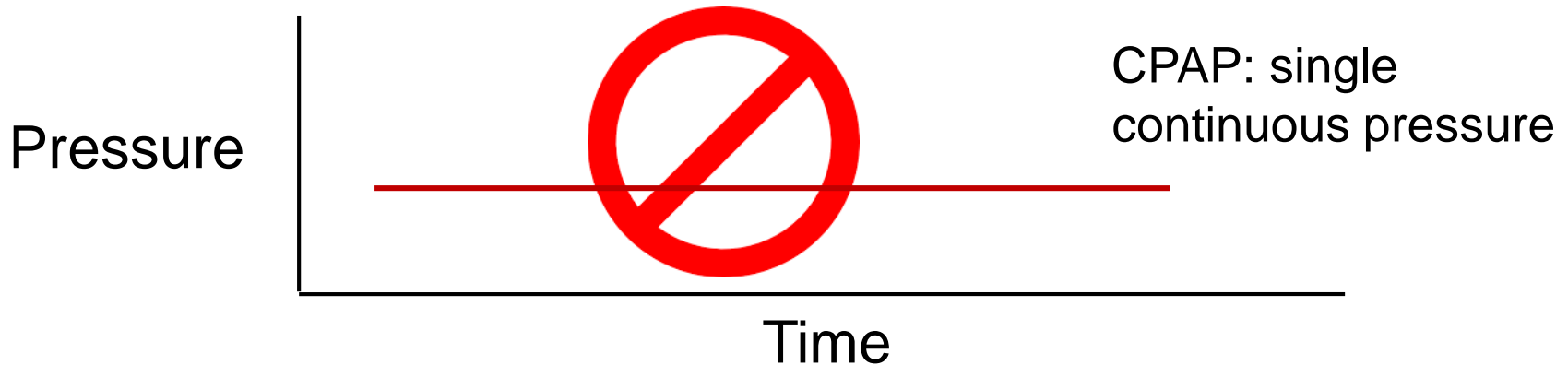
Home mechanical ventilators – non-invasive or invasive



reddot award
product design

cure
SMA

CPAP versus BiPAP



Oxygen without pressure does not help and may hurt you

Positive pressure ventilation

- Goals during **sleep**:
 - Respiratory muscle rest
 - Synchronization
 - Chest wall expansion
- Recommended modes:
 - PC (Pressure control) guaranteed inspiratory time with back up rate
 - ST (spontaneous timed) with back up rate
 - AVAPS (average volume assured pressure support) targeted tidal volume within IPAP range
- Backup respiratory rate required

Non-invasive bi-level positive airway pressure

- IPAP: 14-20 cm of H₂O
- EPAP: 4-6 cm of H₂O
- Respiratory Rate: high enough to capture breathing efforts and rest
- Inspiratory Time: depends on age and set respiratory rate
- Rise time: time between exhalation and rise to peak inspiratory pressure (IPAP)

Invasive bi-level positive airway pressure

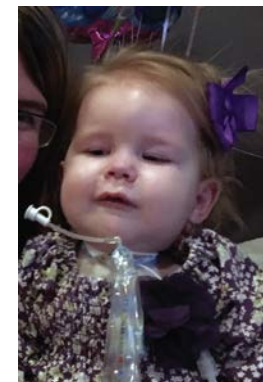
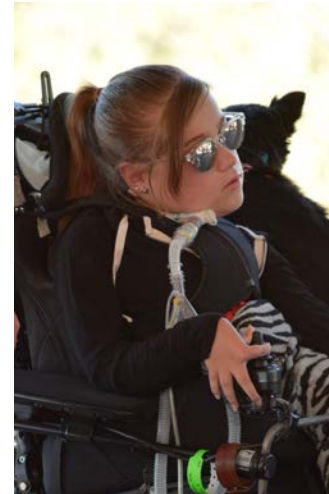
- Ventilation is more efficient
- Modes:
 - Assist control (Every breath is the same)
 - Pressure or volume ventilation
 - Synchronized intermittent mechanical ventilation (SIMV)
 - Pressure or volume ventilation
 - May not be tolerated as well during sleep due to hypoventilation – triggering breath is work
 - AVAPS

Interfaces

Non-invasive



Invasive



Advantages of non-invasive ventilation

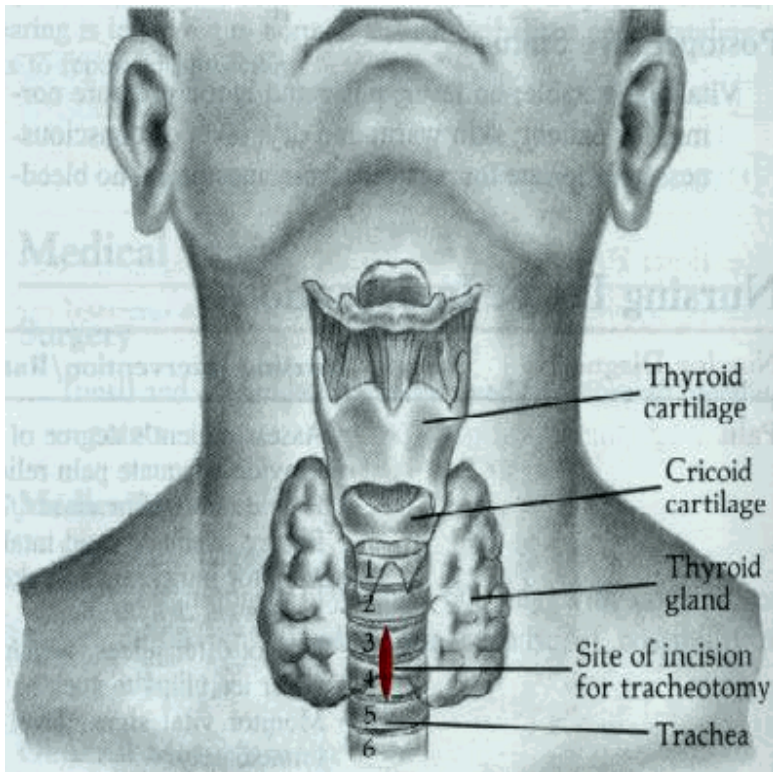
- No surgery
- Reversible
- Safe
- Effective
- Preserves (natural) airway protection mechanisms
- Decreased risk of infection
- Comfortable
- Preserves speech
- Less swallowing issues

Disadvantages of non-invasive ventilation

- Nasal/oral dryness
- Bloody nose
- Nasal congestion
- Sneezing
- Runny nose
- Sinus Infection
- Claustrophobia
- Mask irritation
- Swallowing air
- Mouth leak
- Facial deformities

Invasive ventilation

- Intubation (in an acute illness)
- Tracheostomy with ventilator support – **not** an acute decision



Is a tracheostomy right for me?

PERSONAL/FAMILY CHOICE based on YOUR GOALS

- Non-invasive methods are not effective, tolerated, or manageable in the home
- Excessive oral secretions puts patient at risk for aspiration
- Resources for outpatient management with non-invasive devices are not available
- Failure to extubate despite optimal management

Advantages of tracheostomy

- Bypasses upper airway resistance
- Ventilation is more efficient
- Airway is more protected from aspiration
- Increased ease in suctioning excessive secretions (if needed)
- No mask pressure sores or facial remodeling
- Treats obstructive sleep apnea (if present)
 - In select cases, may be a temporizing measure until extubation is possible
 - Reversible (trach can come out)

Disadvantages of tracheostomy

- Infection
- Bleeding
- Irritation/granulomas
- Accidental dislodgement
- Skin infections – yeast and bacteria
- Need for specialized training and assistance in the home
- Potential for long hospitalization after placement

Tracheostomy Key Points

- Patient and family goals will guide decisions
 - Will a tracheostomy help me/my child reach their/our goals?
- Elective tracheostomy is better than emergency tracheostomy
- The middle of an acute illness is not the best time to be making these types of decisions

Palliative care – not a four letter word

- Recognizes that SMA can be a life threatening disease
- Should be part of the care of the disease process
- Provide symptom relief: pain, shortness of breath, 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 guides how we think and focuses the whole team on goals of care*

Routine pediatric care is part of breathing strong!

- Routine health care
- Immunizations
 - Standard immunizations
 - Influenza vaccination yearly
 - Pneumococcal vaccination
 - Palivizumab (Synagis)
- Proper nutrition
 - Obesity versus malnutrition

What to bring to your appointments

- Your questions and concerns
- All respiratory equipment
 - BiPAP/ventilator
 - Masks
 - Cough assist

Home respiratory equipment

- BiPAP or volume ventilator (when needed)
- Mechanical in-exsufflator (The Cough Assist)
- Suctioning equipment
- Ambu bag
- Pulse oximeter
- Chest cups or high frequency chest wall oscillation (if needed)
- Nebulizer (if needed)

Final message

- 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
- Get involved!

REMEMBER, ANYTHING IS POSSIBLE...

