Respiratory Management in RyR1 Myopathy

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Division of Pediatric Pulmonology
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Sunday, June 24, 2017
Outline

• Progression of Respiratory Disease
• “Timeline” of progression
• Treatment Options
Progression of Disease
Respiratory Failure

Respiratory Muscle Weakness
Respiratory Muscle Weakness

- Diaphragm Weakness
- Chest Wall Muscle Weakness
- Spinal Instability

Respiratory Failure
Respiratory Muscle Weakness

- Diaphragm Weakness
- Chest Wall Muscle Weakness
- Spinal Instability

Respiratory Failure
Respiratory Muscle Weakness

- Chest Wall Muscle Weakness
  - Diaphragm Weakness
  - Chest Wall Stiffness
  - Kyphoscoliosis
    - Low Lung Volumes
      - Increased Respiratory Load
        - Respiratory Failure
  - Spinal Instability
Respiratory Muscle Weakness

- Diaphragm Weakness
  - Poor Airway Clearance
    - Pneumonia
      - Diaphragm Fatigue

- Chest Wall Muscle Weakness
  - Chest Wall Stiffness
  - Low Lung Volumes
    - Increased Respiratory Load

- Spinal Instability

Respiratory Failure
Progression of Disease

- Unaffected
- Fatigue
- Poor Airway Clearance
- Respiratory Failure

Age

Respiratory Capacity
Progression of Disease

- Unaffected
- Fatigue
- Poor Airway Clearance
- Respiratory Failure

Age vs. Respiratory Capacity
Assisted Airway Clearance
Coughing

- Inspiratory
  - Deep breath to get air beyond secretions
  - Stretch expiratory muscles
- Compressive - increased pressure
  - Exhalation against closed glottis
- Expulsive - mobilization
  - Rapid expiratory flow (> 270 L/m)
Support Weak Cough

- Inspiratory Assistance
- Expiratory Assistance
- Inspiratory / Expiratory Assistance
Inspiratory Assistance

- Breath Stacking
- Oronasal Mask / One Way Valve
  - Intact inspiratory muscle strength
- Resuscitation Circuit - Bag and Mask
  - Requires pharyngeal function
- Intermittent Positive Pressure Breaths
Inspiratory Assistance
Expiratory Assistance

- Chest Wall Compression
- Abdominal Compression
- Chest Wall and Abdominal Compression
Expiratory Assistance

Manually Assisted Cough

Rest

Exhalation

Toussaint, et. al. Respir Care 2009
Inspiratory & Expiratory Assistance

- Breath stacking / Manually Assisted Cough
- Mechanical In-Exsufflator (Cough Assist®)
Inspiratory & Expiratory Assistance

- Cough Assist
- Cycle
  - Pi 15-50 cm H$_2$O
  - Apply Pi for 2-3 seconds
  - Pe 15-50 cm H$_2$O 1-2 sec
- 5 cycles - Suction - 5 sets
Table 2—Peak Expiratory Flow Rates During Assisted and Unassisted Coughing

<table>
<thead>
<tr>
<th>Case</th>
<th>Unassisted, L/s</th>
<th>Stack, L/s</th>
<th>Assisted, L/s$</th>
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Mean ± SD 1.81 ± 1.03  3.37 ± 1.07  4.27 ± 1.29  7.47 ± 1.02

Bach, *Chest* 1993
Ventilation
What is Ventilation?

- Bring oxygen into the blood stream
- Remove carbon dioxide produced
  - Maintain acid-base balance
- Cyclic aeration of the lung to remove secretions
Clinical Symptoms of Respiratory Failure

- Morning headaches
- Inadequate Rest
  - Daytime fatigue
  - Failure to thrive
- More rapid loss of function
How to Evaluate Ventilation

- Hypoventilation
  - High Carbon Dioxide
- Night time hypoventilation before daytime
  - First occurs in REM sleep
- Full Sleep Study / 16-Channel Polysomnogram
Sleep Study
### ETCO2 & TCCO2 Summary

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<th>ETCO2 Intervals</th>
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Nocturnal Pulse Oximetry is Not a Viable Alternative
Caution in Diagnosis

- Significant vs. Non-Significant hypercarbia
  - Is EtCO2 > 50 for < 20% sleep time Normal?
- Patients are typically studied when well
  - What about during illness?
- Distinguish hypoventilation from obstructive sleep apnea
How to Support
How not to support

- Supplemental Oxygen
- High-Flow Nasal Cannula Air or Oxygen
- Continuous Positive Airway Pressure (CPAP)
What is Required for Ventilation?

- Interface
  - Nasal
  - Oronasal / Facemask
  - Oral
  - Chest - Negative Pressure Ventilation
- Bi-Level Pressure Generator
Tracheostomy Tube
Interface

• Fit mask to patient’s face
  • Experienced Nurse / Therapist
• Assess for comfort / pressure points
• Desensitization
How to Initiate Ventilation

• Equipment
  • Interface - Nasal / Oronasal
  • Ventilator

• Titration - ideally with patient’s equipment
  • Clinical
  • Sleep Study
Successful Ventilation Initiation

- Patient tolerance
  - Immediate
  - Ongoing
- Parent / Caregiver Training & Competence
- Follow-up to insure adequacy of support
  - May change with clinical status
Ventilation Outcome

Gas Exchange

Mellies, et.al. Eur Respir J. 2003
Use of a Tracheostomy Tube

- **Absolute Indication**
  - Incompetent upper airway / inability to be extubated

- **Relative indications for tracheostomy tube**
  - Interface problems - skin integrity
  - Facial interface refusal / intolerance
  - Duration of NIPPV is not an absolute indication
  - Alter interfaces
Progression of Disease

- Adjust level and amount of NIPPV support based on patient’s need
- No advantage to more NIPPV support (duration or pressure) than is needed
- Be aware that normal ventilation when well does not insure normal ventilation when acutely ill
Progression of Disease

- Unaffected
- Fatigue
- Poor Airway Clearance
- Respiratory Failure
- Respiratory Capacity

Age
Acute Illness

Age

Unaffected

Fatigue

Poor Airway Clearance

Respiratory Failure

Respiratory Capacity
Conclusion

• The onset of respiratory symptoms can be subtle

• Because the first onset can occur during an acute illness it is important to be prepared and proactive

• There are a variety of effective options to support airway clearance and ventilation

• Treatment needs to be fit to the patient not the patient fit to the treatment
mayero@email.chop.edu
Clinical Titration

• Normalize CO2
  • Continuous EtCO2 / TcCO2 at night
• Chest Wall Motion - Maximize excursion
• Symptom Improvement
  • Quality of Sleep
  • Morning Headaches
Sleep Study Titration

• Titrate Pi and Pe to
  • Improve hypercapnea / oxygenation
  • Improve work of breathing
    • Thoracoabdominal asynchonry
  • Diagnosis & titration may not be feasible together
Physiologic Titration

- Reduce EtCO2 by 5% with maximal Pi and Pe
- Reduce work of breathing using Pi and Pe

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<thead>
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<th>TABLE 3. SLEEP DATA DURING MECHANICAL VENTILATION WITH THE TWO SETTINGS</th>
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<tr>
<td>TST, min</td>
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<td>SE, % of TST</td>
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<td>SWS, % of TST</td>
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<td>REM, % of TST</td>
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<td>Arousal index, events/h</td>
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<td>ODI, events/h</td>
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<tr>
<td>SaO2 nadir, %</td>
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<td>TST90, %</td>
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<td>NREM ineffective efforts, events/h</td>
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*Definition of abbreviations: NREM = non-REM sleep; ODI = oxygen desaturation index (number of desaturation episodes · h of sleep or time in bed); PHYS = physiological setting; SE = sleep efficiency; SWS = slow wave sleep; TST = total sleep time; TST90 = time spent with SaO2 < 90% (% of total sleep time or time in bed); US = usual setting.*

Fanfulla, et. al. Am J Respir Crit Care Med 2005
NIPPV Outcomes

- Acute Care
  - Successful support in 12 of 16 cases
  - NIPPV and aggressive Cough Assist treatment

Inspiratory Assistance

Dohna-Schwake, et.al. Pediatr Pulmonol
2006
Preventative Management
Preventative Management

- Immunizations
  - Keep up to date with standard schedule
- Seasonal Influenza Vaccination
  - No evidence for Palivizumab (RSV vaccine)
- Respiratory Range of Motion
Range of Motion

• Extend motion around joint to maximal

• All directions

• Hold at point of maximal extension/flexion

• Duration

• Repetition
# Hyperinsufflation

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<th>Major Diagnoses</th>
<th>No. of Patients</th>
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<th>Mean VCsit</th>
<th>Mean MIC</th>
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<td>DMD</td>
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<td>622 (1-2710) ± 595</td>
<td>1252 (220-3280) ± 670</td>
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<td>Myotonic</td>
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<td>1131 (0-3580) ± 744</td>
<td>1712 (30-5100) ± 926</td>
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Respiratory Muscle Weakness

- Spinal Instability
- Chest Wall Muscle Weakness
  - Increased Respiratory Load
  - Low Lung Volumes
  - Diaphragm Fatigue
  - Pneumonia
  - Poor Airway Clearance
  - Chest Wall Dysfunction

- Kyphoscoliosis

Muscle Intervention

Clinical Intervention

Respiratory Failure
### Table 2—Peak Expiratory Flow Rates During Assisted and Unassisted Coughing

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<tr>
<td>12</td>
<td>1.66</td>
<td>3.14†</td>
<td>4.62</td>
<td>8.14</td>
</tr>
<tr>
<td>13</td>
<td>4.02</td>
<td>4.44†</td>
<td>4.77</td>
<td>7.46</td>
</tr>
<tr>
<td>14</td>
<td>2.33</td>
<td>4.50†</td>
<td>4.37</td>
<td>7.48</td>
</tr>
<tr>
<td>15</td>
<td>3.34</td>
<td>4.70†</td>
<td>4.92</td>
<td>7.78</td>
</tr>
<tr>
<td>16</td>
<td>0.97</td>
<td>1.78†</td>
<td>2.58</td>
<td>8.17</td>
</tr>
<tr>
<td>17</td>
<td>1.02</td>
<td>1.48†</td>
<td>3.32</td>
<td>6.42</td>
</tr>
<tr>
<td>18</td>
<td>1.00</td>
<td>2.25†</td>
<td>2.25</td>
<td>9.23</td>
</tr>
<tr>
<td>19</td>
<td>1.50</td>
<td>2.40†</td>
<td>4.40</td>
<td>7.80</td>
</tr>
<tr>
<td>20</td>
<td>1.51</td>
<td>4.33*</td>
<td>6.33</td>
<td>9.11</td>
</tr>
<tr>
<td>21</td>
<td>1.58</td>
<td>3.88*</td>
<td>3.68</td>
<td>7.81</td>
</tr>
</tbody>
</table>

**Mean ± SD**

- Unassisted: $1.81 ± 1.03$
- Stack: $3.37 ± 1.07$
- Assisted: $4.27 ± 1.29$
- Exsufflator: $7.47 ± 1.02$

Bach, Chest 1993
Inspiratory & Expiratory Assistance

Toussaint, et. al. Respir Care 2009

270 L/m
Inspiratory Assistance

- Intermittent Positive Pressure Breathing
  - Pressure with maximal inspiratory volume
  - Hold volume for 1-2 Seconds
Intrapulmonary Percussive Ventilation

- High frequency oscillation under pressure
- Nebulization during treatment
- Performed during tidal breathing
Intrapulmonary Percussive Ventilation

Exhalation without PercussiveTech HF

Exhalation with PercussiveTech HF
NIPPV Outcome

Gas Exchange

Mellies, et.al. Eur Respir J. 2003
NIPPV Outcome

Gas Exchange

Mellies, et.al. Eur Respir J. 2003
NIPPV Outcomes

Sleep Quality

Table 2 – Influence of noninvasive ventilation (NIV) on sleep-disordered breathing and sleep

<table>
<thead>
<tr>
<th></th>
<th>Before NIV</th>
<th>During NIV</th>
<th>p-value</th>
</tr>
</thead>
<tbody>
<tr>
<td>RDI·h⁻¹</td>
<td>10.5±13.1</td>
<td>3.1±3.5</td>
<td>&lt;0.001</td>
</tr>
<tr>
<td>REM-RDI·h⁻¹</td>
<td>20.5±21.1</td>
<td>3.0±5.3</td>
<td>&lt;0.001</td>
</tr>
<tr>
<td>Arousal index·h⁻¹</td>
<td>20.6±14.3</td>
<td>10.2±3.8</td>
<td>&lt;0.001</td>
</tr>
<tr>
<td>Light-sleep %</td>
<td>55±12</td>
<td>44±13</td>
<td>&lt;0.05</td>
</tr>
<tr>
<td>Slow-wave-sleep %</td>
<td>24±9</td>
<td>34±9</td>
<td>&lt;0.05</td>
</tr>
<tr>
<td>REM-sleep %</td>
<td>18±6</td>
<td>20±6</td>
<td>0.18</td>
</tr>
</tbody>
</table>

Mellies, et.al. Eur Respir J. 2003
Two Approaches To Treatment
Two Approaches

• Reactive
  • Treat symptoms after they have occurred and are established

• Proactive / Preventative
  • Treat symptoms as early as possible as they are starting
  • Prevent symptoms from occurring at all
Reactive - Unaffected

- Cold symptoms start
- Nasal congestion
- Fever
- Cough
- Medication
- Resolution
Reactive - DMD

- Cold symptoms start
- Nasal congestion
- Fever
- Cough
- Medication
- Resolution
Reactive - DMD

- Cold symptoms start
- Nasal congestion
- Fever
- Cough
- Medication
- Resolution
- Poor cough
- Prolonged illness
- +/- Pneumonia
Proactive Approach

- Cold symptoms start
- Nasal congestion
Proactive Approach

- Cold symptoms start
- Nasal congestion
- Fever
- Cough
- Medication
- Resolution

Assisted Airway Clearance
Preventative Management

• Immunizations
  • Keep up to date with standard schedule
  • Seasonal Influenza Vaccination
  • No evidence for Palivizumab (RSV vaccine)

• Respiratory Range of Motion - Dr. Sawnani
Proactive Management

• Cough a lot and cough early
• Insuring an effective cough
• Supporting an ineffective cough
Coughing

- Inspiratory
  - Deep breath to get air beyond secretions
  - Optimize length tension relationship of expiratory muscles

- Compressive - increased pressure
  - Exhalation against closed glottis

- Expulsive - mobilization
  - Rapid expiratory flow (> 160-180 L/m)
Augment Weak Cough

- Inspiratory Assistance
- Expiratory Assistance
- Inspiratory / Expiratory Assistance
- Other
Inspiratory Assistance
Inspiratory Assistance

• Intermittent Positive Pressure Breathing
  • Pressure with maximal inspiratory volume
  • Hold volume for 1-2 Seconds
  • Exhalation at maximal capacity of patient
Inspiratory Assistance

Dohna-Schwake *Pediatr Pulmonol* 2006
Inspiratory Assistance

- Breath Stacking
- Oronasal Mask / One Way Valve
- Intact Inspiratory Muscle Strength
- Intermittent Positive Pressure Breaths
Inspiratory Assistance

- Chest Wall Compression
- Abdominal Compression
- Chest Wall and Abdominal Compression
Expiratory Assistance

Manually Assisted Cough

Rest

Exhalation

Toussaint, et. al. Respir Care. 2009.
Inspiratory & Expiratory Assistance

• Breath stacking / Manually Assisted Cough
• Mechanical In-Exsufflator (Cough Assist®)
## Inspiratory & Expiratory Assistance

<table>
<thead>
<tr>
<th>Major Diagnoses</th>
<th>No. of Patients</th>
<th>Mean Age</th>
<th>Mean CPF</th>
<th>Mean ACPF</th>
</tr>
</thead>
<tbody>
<tr>
<td>DMD</td>
<td>53</td>
<td>26 (14–44)</td>
<td>1.58 (0.1–5.7) ± 1.7</td>
<td>3.76 (0–6.2) ± 1.4</td>
</tr>
<tr>
<td>Myotonic</td>
<td>6</td>
<td>47 (36–53)</td>
<td>4.03 (2.7–4.7) ± 0.8</td>
<td>5.10 (4.4–5.9) ± 0.6</td>
</tr>
<tr>
<td>Other myopathies</td>
<td>55</td>
<td>39 (11–85)</td>
<td>3.11 (0.1–7.5) ± 1.8</td>
<td>4.64 (1.6–7.5) ± 1.4</td>
</tr>
<tr>
<td>SMA</td>
<td>31</td>
<td>19 (7–56)</td>
<td>1.76 (0.1–4.6) ± 1.5</td>
<td>3.54 (1.7–5.5) ± 1.1</td>
</tr>
<tr>
<td><strong>Total</strong></td>
<td><strong>282</strong></td>
<td><strong>42 (7–85) ± 20</strong></td>
<td><strong>2.52 (0.1–9.7) ± 2.0</strong></td>
<td><strong>4.28 (0–9.7) ± 2.2</strong></td>
</tr>
</tbody>
</table>
Inspiratory & Expiratory Assistance

Toussaint, et. al. Respir Care. 2009.
Cough Assist
Inspiratory & Expiratory Assistance

Cough Assist Cycle

Pi 15-50 cm H₂O
Hold at Target Pi for 1-2 seconds
Pe 15-50 cm H₂O 1-2 sec

5 cycles
Suction
5 sets
Inspiratory & Expiratory Assistance

Fauroux *Chest* 2008
Intrapulmonary Percussive Ventilation

- High frequency oscillation under pressure
- Nebulization during treatment
- Performed during tidal breathing
Intrapulmonary Percussive Ventilation
Respiratory Muscle Weakness

Chest Wall Muscle Weakness

Diaphragm Weakness

Poor Airway Clearance

Pneumonia

Spinal Instability

Muscle Intervention

Clinical Intervention
Intermittent Positive Pressure Breaths (IPPB)

- Apply inspiratory pressure
- Hyperinflate lungs
- Increase airway caliber
- Passive recoil
- Can be used with nebulization