MYOTONIC DYSTROPHY
PULMONARY CONSIDERATIONS:
SCIENCE & MANAGEMENT STRATEGIES

“When you can’t breathe nothing else matters”
American Lung Association
Overview

- Biology refresher course
  - The lungs and what they do
- What goes wrong in myotonic dystrophy
- Evaluating breathing problems
- Evaluating sleep problems
- Treatment options
  - Lung recruitment
  - Ventilation
  - Secretion clearance
  - Sleepiness
  - Emergency plans
Why Should I Care About This?

- Respiratory failure is a leading cause of death in myotonic dystrophy

One day his mother said when Pierre climbed out of bed, “Good morning, darling boy, you are my only joy.” Pierre said, “I don't care!”
Respiratory Muscles

- **Inspiratory**: muscles increase thoracic volume, intrapleural and alveolar pressures fall. Air is drawn in to lungs.
  - Diaphragm
  - External intercostals
  - Scalene, sternocleidomastoid

- **Expiratory**: Largely passive
  - Abdominal muscles
  - Internal intercostals
The Lung’s Main Function

- Get Oxygen into the body & blood stream
  - Blood carrying oxygen goes to the heart
  - Heart pumps it to all the organs

- Get rid of carbon dioxide
  - Our bodies produce carbon dioxide (CO2)
  - When we exhale, we blow out carbon dioxide
Control of Ventilation (Breathing)
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- **Major Mechanism:**

  - Sensors in the brainstem respond to carbon dioxide levels.
Control of Ventilation

- Minor mechanism:
  - sensors respond to low oxygen levels

- Some people with DM don’t respond normally to high carbon dioxide

- Some people with DM stop breathing when given too much oxygen
Oxygen Can Be Dangerous

- People adjusted to elevated carbon dioxide may stop breathing when given high levels of oxygen.

- People with DM should not receive oxygen without ventilatory support or close monitoring.
Respiratory Complications Are a Major Cause of Morbidity and Mortality in Neuromuscular Disorders

Respiratory Complications in MD

- Complications Includes
  - Pneumonia
  - Respiratory Failure (elevated CO2 or low oxygen)
  - Atelectasis (collapse of air sacks)
9 adults with Myotonic Dystrophy

- 6 of 9 had respiratory symptoms – mild shortness of breath
- 7 had mild to moderate abnormalities in pulmonary function
  - Weak breathing muscles
- Arterial carbon dioxide was elevated to 50 (normal < 44)
- Blunted response to breathing carbon dioxide (0.79L/min/mmHgCO₂ vs. 1.2)
- Breathing 100% oxygen slowed breathing and raised carbon dioxide
General Considerations

- Respiratory complications are a frequent cause of death in DM
  - Can contribute to sleepiness
- Many DM patients do not get routine pulmonary evaluation and testing
- Respiratory equipment is underutilized
- Sleep can be abnormal in DM
- Anesthesia can be problematic in DM
The Pulmonologist Visit
What can I expect during a pulmonary visit?

- Interview about pulmonary problems and symptoms
- Physical examination
  - Look at respiratory muscle use
  - Listen to lungs
- Pulmonary function tests
- Discuss respiratory treatments
- Discuss advanced directives
- Discuss research
What symptoms should I look for?

- Shortness of breath
  - Especially when lying supine
- Difficulty sleeping
- Daytime sleepiness/fatigue
- New headaches
- Inability to take deep breath or cough
- “Chest Congestion”
- Fevers, chest pain, phlegm
The Pulmonary Evaluation

- Pulmonary Function Testing
  - Can follow disease course
  - Diagnose respiratory failure
  - Help timing of interventions
    - Ventilatory support
    - Feeding tubes
    - Oxygen (mostly for air travel)
Spirometry: The Forced Vital Capacity (FVC)

FVC is Usually Reported as % Predicted Value
Pitfalls of Pulmonary Testing

- Tests require a tight seal around a mouthpiece
- Weakness of facial muscles in DM can be a problem
Other pulmonary tests

- Lung Volumes / Diffusing capacity
- Oximetry
- Capnography
  - measure of exhaled carbon dioxide
- Arterial blood gas
- Maximal inspiratory pressure
- X-Rays
- Sleep Study
Sleep Problems
Terminology of Sleep Apnea

- Obstructive Events: ‘Cannot Breath’
- ‘Mixed’
- Central Events: ‘Won’t Breath’
- Reduced Drive
Neuromuscular Activity and Upper Airway Obstruction

UPRIGHT

AIRWAY OPEN

Genioglossal EMG: INSPIRATORY BURSTS

SUPINE

TENDENCY OF TONGUE TO RELAPSE

AIRWAY OPEN

Genioglossal EMG: TONIC ACTIVITY + INSPIRATORY BURSTS

SUPINE

TONGUE RELAPSED TOWARD POSTERIOR PHARYNGEAL WALL

INCREASED AIRWAY RESISTANCE

Genioglossal EMG: GREATLY DIMINISHED OR ABSENT ACTIVITY
Clinical Aspects: Diagnosis of Sleep Apnea

- Polysomnography: Standard diagnostic test
  - Electro-oculogram (EOG)
  - Electroencephalogram (EEG)
  - Electromyogram (EMG)
  - Electrocardiogram (ECG)
  - Airflow
  - Chest and Abdominal Wall Effort
  - Body Position
  - Oxyhemoglobin Saturation

- Recording time ~ 6 to 8 hours
Tracing of Normal Respiration During Sleep

- EOG (L)
- EOG (R)
- EMG (S)
- C3-A2
- C3-O1
- EKG
- EMG (L)
- SaO2
- Airflow (Cannula)
- Airflow (Thermistor)
- Thoracic
- Abdomen
Assessing Need for Assisted Cough

- PCF of at least 160 L/min is necessary for the successful extubation or tracheostomy tube decannulation of patients with neuromuscular disease
- PCF decreased during infections (270 L/min cutoff)

Chest Volume 110, Issue 6, 1996, Pages 1566-1571
Treatment of Breathing Problems
High Frequency Chest Wall Oscillation (HFCWO) – “The Vest”

- Vibration and chest compression loosen mucus and make it easier to cough out
- Recent study showed lower health care costs after getting a vest
- Can be used in conjunction with cough machine
Mechanical In-Exsufflation or Cough Assist

- Applies positive (+) pressure to the airways then rapidly shifts to a negative (-) pressure
- Can generate peak cough flow of 300-660 L/min to simulate a cough
- Delivered via mask, mouthpiece, trach tube
Cough Assist – When?

- 2-4 sessions per day
  - 1 session = 3-5 treatments
  - 1 treatment = 3 to 5 cycles
  - 1 cycle = 1 insufflation and 1 exsufflation

- Best before meals and at bedtime

- Can also use any time secretions are present

- Avoid hyperventilation – no more than every 10 minutes
Cough Assist Video
Non-Invasive Ventilation (BiPAP)

- Delivers positive pressure
  - Inflates lungs
- Start using at night
- Can use during day as needed
  - Some use ~24hrs a day
Non-Invasive Ventilation: Benefits

- Expand lungs/prevent atelectasis
- Preserve thoracic range of motion
- Decrease shortness of breath
- Improve sleep quality
- Improve “Quality of Life”
- Improve cognitive function
- Prolong survival
  - Greater effect than any medication or other treatment
Non-Invasive Ventilation: Concerns

- Finding correct mask is important
Masks and Myotonic Dystrophy

- Facial weakness can make mask fit a challenge
Non-Invasive Ventilation: Concerns

- Most Common Problems:
  - Skin irritation/breakdown
  - Nasal congestion or drainage
  - Claustrophobia / not comfortable with mask and BiPAP

- Common Solutions
  - Different mask / alternate masks
  - Heat & humidify air
  - Decongestants / nasal sprays
  - Gradually increase use and pressure of BiPAP
Non-Invasive Ventilation: Questions

- When should BiPAP be started?
  - Current guidelines and reimbursement
    - FVC < 50%, Oxygen sat < 88% for 5 minutes, MIP > -60, PaCO2 > 45
  - May have benefit at earlier times
  - May be better to target symptoms
Sleepiness & Fatigue

- A common symptom in DM
- Can be due to sleep problems
  - Though some with sleep problems don’t feel tired and some who are tired don’t have sleep problems
- Can be due to breathing problems
- If sleepiness persists with treatment of breathing and sleep issues consider modafanil (Provigil)
Surgery

- Anesthesia can cause prolonged effects in DM
- Can lead to delayed recovery after surgery
- Can lead to low oxygen levels after surgery
- Need to be sure anesthesiologists are knowledgeable about DM
  - Opioid pain meds should be limited
  - Succinylcholine should be avoided
  - Need close monitoring post-op
Prevention

- Avoiding influenza and respiratory infections is important
  - Get yearly flu vaccine
- Enforce hand hygiene
- Avoid people who are sick
Emergency Preparedness

- www.ventusers.org
- Home vent users emergency checklist
- Caregiver’s Emergency Preparation checklist
- Patients Vital Information for Medical Staff
- Treating Neuromuscular Patients Who Use Home Ventilation: Critical Issues

- Patients and family members often know more about DM than providers
  - Need to be an advocate
Case 1: 37yo woman

- Has normal lung function, is active
- Has heart block, arrhythmias and has a pacemaker
- Had surgery for benign abdominal mass
  - Plan was to go home same day
- Post-op had shortness of breath and low oxygen saturation
- Required overnight ICU monitoring and oxygen
- Did well after 36 hours
Case 2: 40 yo man

- Diagnosed at age 40 with trouble gripping tennis racket
- Had sleepiness and had sleep study
  - Mild to moderate apnea
  - Started CPAP
- Felt better – but ....
Case 2 continued

- Checked Pulmonary Function
  - Vital capacity = 30% predicted
  - Carbon dioxide = 52 (normal < 45)
- Changed CPAP to BiPAP
- Condition has been relatively stable for several years
Summary

- Breathing involvement in DM is highly variable
- Weakness of inspiratory and expiratory muscles is common
- Illness and death due to breathing complications occurs
- Oxygen can cause problems – be cautious
- Routine pulmonary evaluation is important
- Treatment may include CPAP, BiPAP, assisted cough
- Sleep evaluation is also important
Questions