

## Breathing Health in FSHD

Joshua O. Benditt, MD  
Medical Director, Respiratory Care Services  
University of Washington Medical Center

---

---

---

---

---

---

---

---

## Topics

- Pertinent anatomy
- The problem with sleep and neuromuscular diseases
- Measurement of respiratory function in clinic
- Noninvasive techniques for support
- What you can do to maintain a healthy respiratory system

---

---

---

---

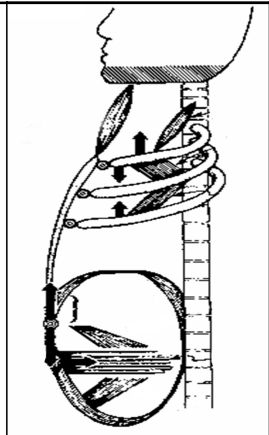
---

---

---

---

## Breathing Muscles



---

---

---

---

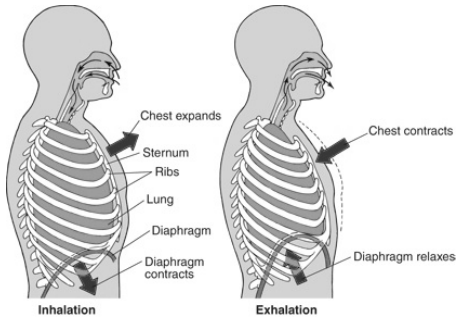
---

---

---

---

## Breathing Movement



---

---

---

---

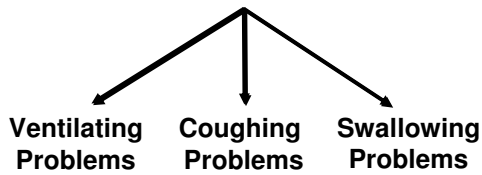
---

---

---

---

## Neuromuscular Respiratory Problems



---

---

---

---

---

---

---

---

## FSHD and Breathing Issues

- Followed 53 patients with FSHD over 10 years.
- 50% of the patients had abnormalities on breathing measurements
  - 13 % had severe involvement
  - 22% had some pulmonary complications
    - Kilmer Det al. Profiles of neuromuscular diseases: facioscapulohumeral muscular dystrophy. *Am J Phys Med Rehabil* 1995;74(Suppl); 131-139.
- More recent study in adults
  - Smaller percentage had breathing problems
  - Patients were adults who may have had a less severe form of disease.
    - Wolgemuth M, van der Kooi EL, Van Kesteren RG, et al. Ventilatory support in facioscapulohumeral muscular dystrophy. *Neurology* 2004;63:176-178.

---

---

---

---

---

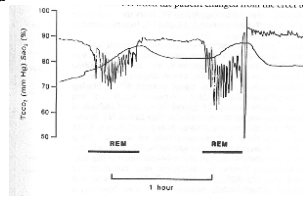
---

---

---

## Ventilating Problems and Sleep

- During sleep, particularly REM, central output to the respiratory muscles decreases
- Weak muscles + decreased drive results in significant hypoventilation with CO<sub>2</sub> retention
- Can result in daytime hypercarbia



• Bye et al. Thorax 1990;45:241-247.

---

---

---

---

---

---

---

---

## Symptoms of Sleep Problems

- Unexplained nocturnal awakenings
- Vivid nightmares
- Unexplained nightsweats
- Morning headaches
- Daytime hypersomnolence
- Associated:
  - changes in concentration
  - hypertension
  - depression

---

---

---

---

---

---

---

---

## Ventilatory Assessment

- **Forced Vital Capacity**
  - Volume of the biggest breath you can take.
  - supine (if diaphragm weakness suspected)
- **Maximal Inspiratory Pressure (MIP)**
  - Breath in as hard as you can against a pressure measuring device
- **Carbon dioxide**
  - Arterial blood gas
  - End-tidal (exhaled breath)
  - Transcutaneous

---

---

---

---

---

---

---

---

### **Cough Problems**

- Abdominal and rib muscle weakness
- Inability to close glottis
- Reduced peak cough airway flow
- Inability to clear secretions
- Mucous build-up and infection

---

---

---

---

---

---

---

---

### **Expiratory (Cough) Failure Assessment**

- Peak Cough Flow
  - Normal > 500 lpm
  - < 160 lpm ineffective clearance
  - < 270 lpm worrisome
    - drops to < 160 seen during infection
- Maximal expiratory pressure
  - ? Minimal value predictive of problems

---

---

---

---

---

---

---

---

### **Cough Peak Flow Meter**



---

---

---

---

---

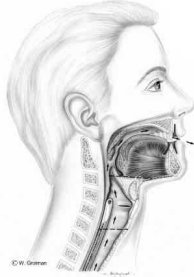
---

---

---

## Glottic and Swallowing Muscles

- Muscles of mastication
- Tongue
- Pharyngeal and hypopharyngeal muscles
- Laryngeal muscles



---

---

---

---

---

---

---

---

## Glottic Failure

Airway Protection, Cough and Swallowing

- Intimately related to cough function
- Bulbar muscle involvement
- Portends poorer prognosis
- Difficulty in managing secretions
- Choking episodes and aspiration
- Pneumonia and respiratory failure

---

---

---

---

---

---

---

---

## Measurement of Swallowing Function

- At bedside or clinic
  - history
  - swallowing challenge
- Barium swallowing study
- Direct endoscopic visualization

---

---

---

---

---

---

---

---

## Goals of Management

- Ameliorate symptoms
- Improve sleep quality
- Improve quality of life
- Improve and stabilize gas exchange
- Extend survival

---

---

---

---

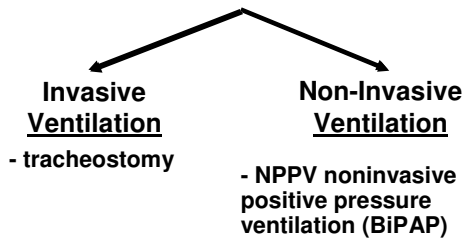
---

---

---

---

## Treatment for Ventilatory Problems



---

---

---

---

---

---

---

---

## Interfaces for NPPV-Nocturnal



---

---

---

---

---

---

---

---

## Nocturnal Ventilators

- Bilevel (pressure support)
- Back-up rate
  - Hypopneas and apneas invariably present
- Modern devices allow for adjustment of inspiratory flow rates
  - “rise time”
- Humidification is important



---

---

---

---

---

---

---

---

## Initiating Nocturnal NPPV

- Screen for symptoms
- Sleep study diagnostic
- or FVC < 50% predicted
- or MIP > -60 cm water pressure
- or PaCO<sub>2</sub> > 45 mm Hg
- Noninvasive ventilation
  - BiPAP or VPAP
  - Nasal mask
  - Mouth leak

---

---

---

---

---

---

---

---

## 24 Hour Per Day NPPV Combination Therapies

- Nocturnal ventilation
  - nasal or oronasal ventilation with pressure or volume
  - mouthpiece ventilation (Europe)
- Daytime support
  - If needed
    - dyspnea during day
    - PaCO<sub>2</sub> remains elevated despite nocturnal therapy
  - mouthpiece ventilation
  - occasionally nasal ventilation (ALS)
- Oxygen is almost never needed and can be detrimental

---

---

---

---

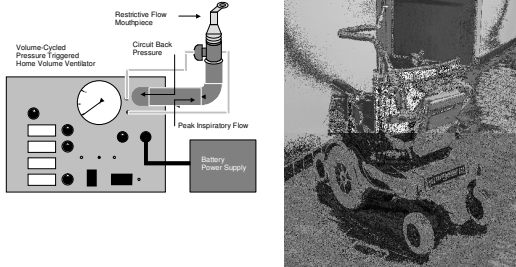
---

---

---

---

## Mouthpiece Ventilator Set-up ("Sip Ventilation")




---



---



---



---



---



---



---

## When Must Tracheostomy Be Considered ?

- Significant glottic dysfunction
- Elevated PaCO<sub>2</sub> despite optimal noninvasive therapy
- Recurrent pneumonia
- Patient preference
- Lack of experienced healthcare providers for NPPV

---



---



---



---



---



---



---

## Expiratory or Cough Assistance

- Cough augmentation
  - Assisted cough
    - Abdominal or lateral ribcage thrust
    - Postural maneuvers
  - Resuscitation bag
    - breath stacking
    - elastic recoil augments cough function
  - Cough-Assist (Inexsufflator)
    - requires relatively intact bulbar function
  - Mouthpiece ventilator
    - breath stacking also possible

---



---



---



---



---



---



---



## Cough Augmentation Mechanical Inexsufflation (Cough Assist™)




---

---

---

---

---

---

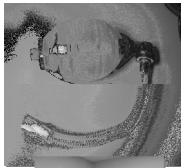
---

---

## Manual & Mechanical Cough Augmentation

### Manual / Mechanical hyperinflation:

- Increased expiratory flow volume and velocity
- Intrathoracic pressure
- Portable therapy
- Oral strength limitation
- Skilled caregiver




---

---

---

---

---

---

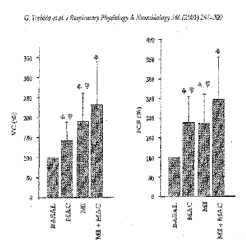
---

---

## Mechanical + Manual Cough Augmentation

### Mechanical Ventilation Supported Insufflation + Manual Assisted Cough

Trebbia G, et al, *Resp Phys & Neurobiol*, 146 (2005) 291-300




---

---

---

---

---

---

---

---

## Treatment of Swallowing Problems

- Head down
- Thick liquids
- Avoidance of dry, bulky foods
- Secretion management
- Tracheostomy
- Gastrostomy tube

---

---

---

---

---

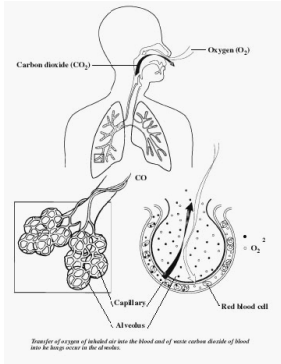
---

---

---

## Oxygen and Carbon Dioxide

- As carbon dioxide goes up, oxygen levels go down
- Weak muscles can cause carbon dioxide to go up
- Giving oxygen when carbon dioxide is up can blunt respiratory drive making CO<sub>2</sub> go even higher
- Pulse oximeter finger probes do not measure carbon dioxide but do measure oxygen levels
- If you are sick with respiratory problems in the ER blood gas should be checked not just pulse oximeter




---

---

---

---

---

---

---

---

## Maintaining Your Respiratory Health

- Avoiding infection
  - Flu shot, pneumonia shot
  - Avoid sick individuals during peak infection season
  - Use hand sanitizer
- Look for symptoms:
  - Of sleep problems
  - Recurrent infections or weak cough
- Assessment
  - Annual vital capacity
  - Other tests as indicated
- ER visit for respiratory infection
  - Oxygen may be given but a check of carbon dioxide must be undertaken
  - Oxygen blunts drive to breathe and may worsen elevated carbon dioxide level

---

---

---

---

---

---

---

---