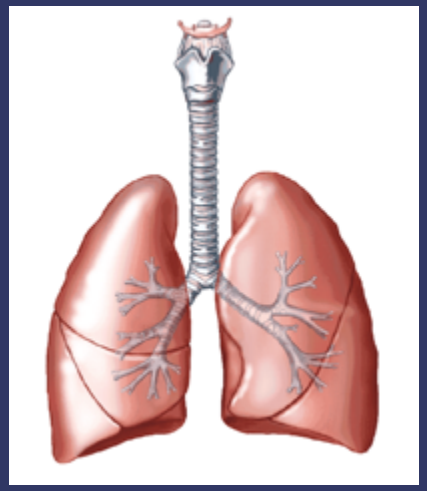
# MYOTONIC DYSTROPHY PULMONARY CONSIDERATIONS: SCIENCE & MANAGEMENT STRATEGIES



"When you can't breathe nothing else matters"
American Lung Association

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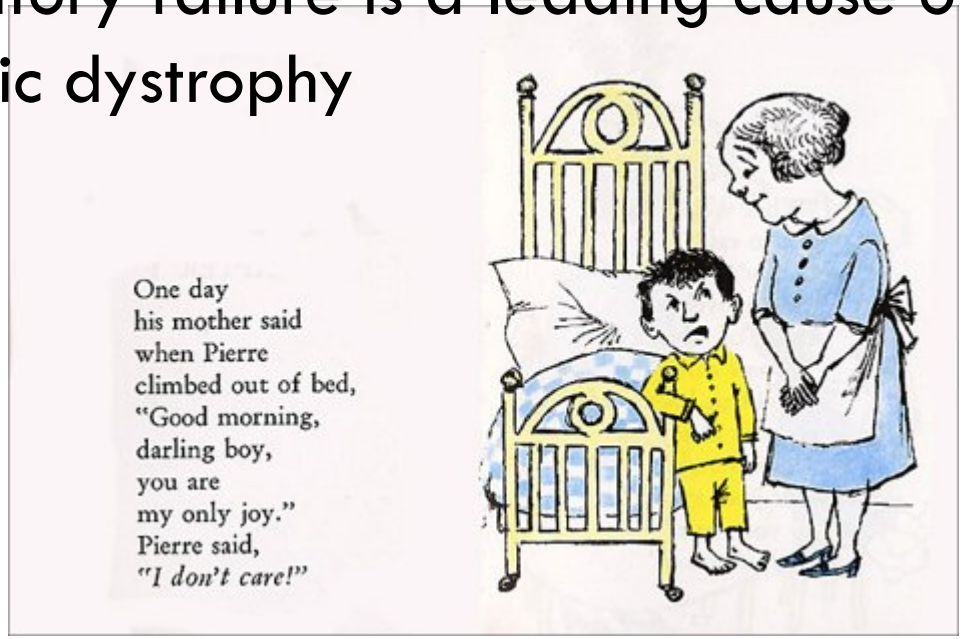
# Overview

- 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

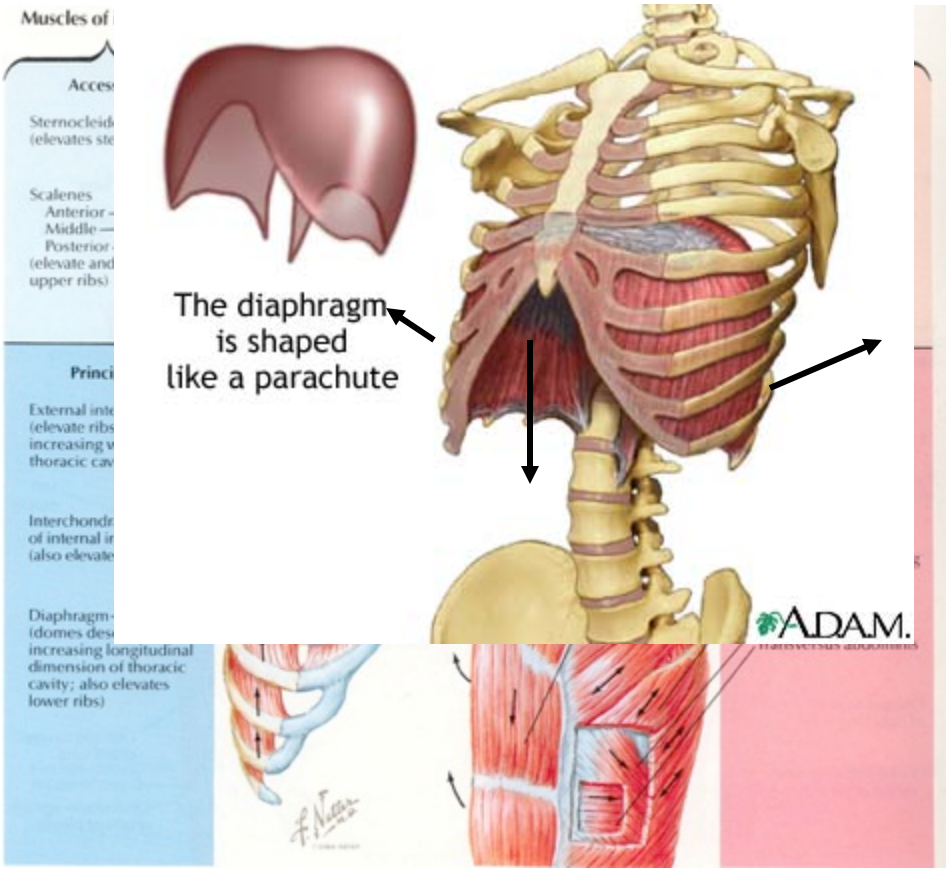
# Why Should I Care About This?

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



# 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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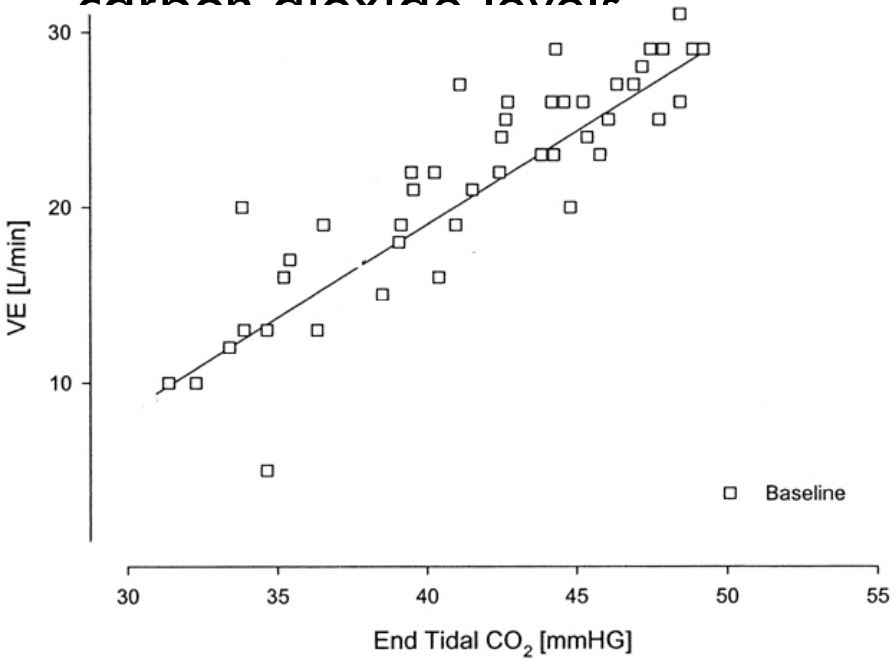
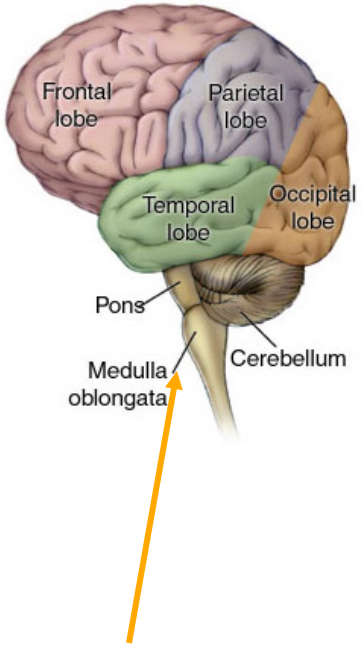
- Major Mechanism:
  - Sensors in the brainstem respond to carbon dioxide levels



# Control of Ventilation

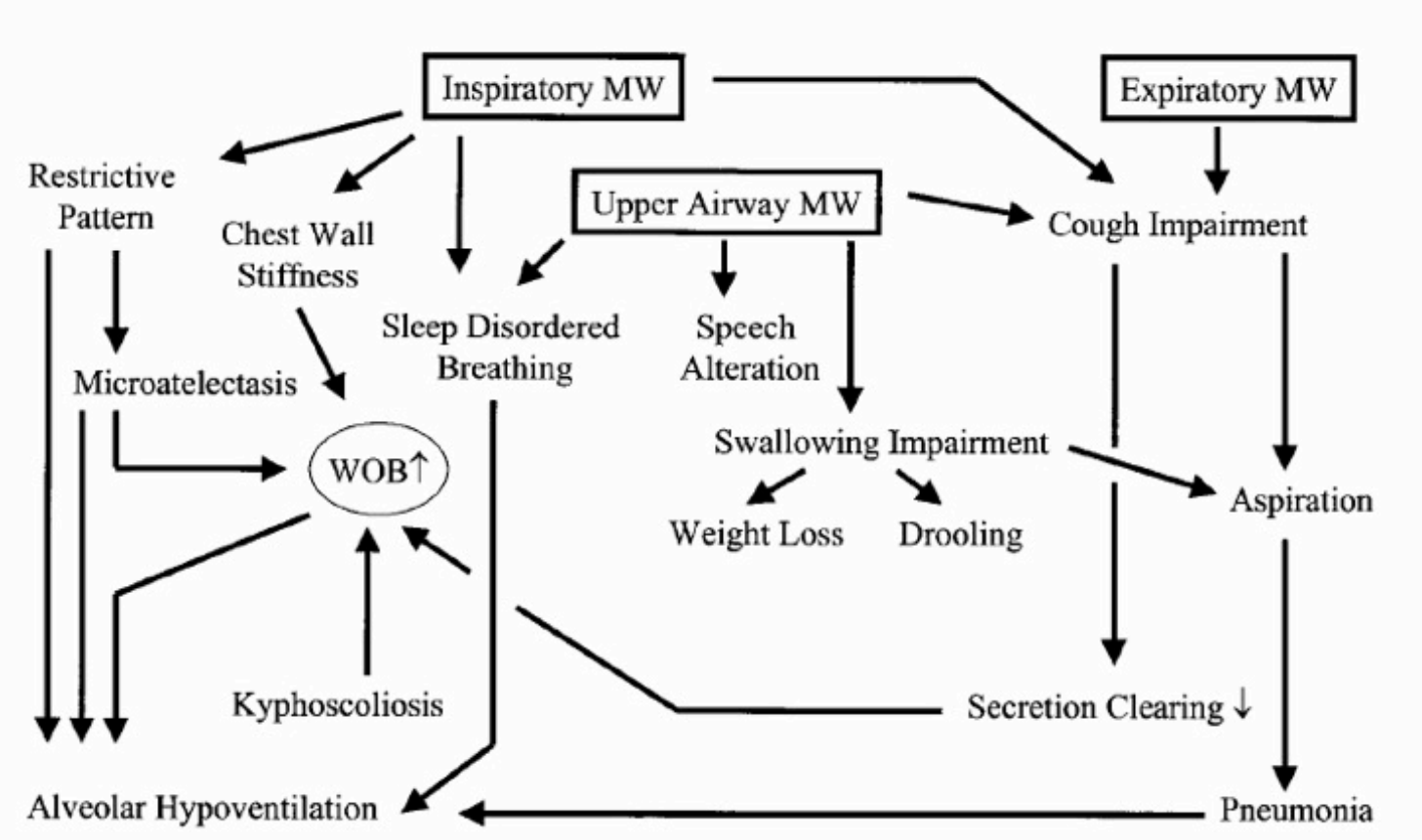
- Minor mechanism:
  - sensors respond to low oxygen levels

- 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



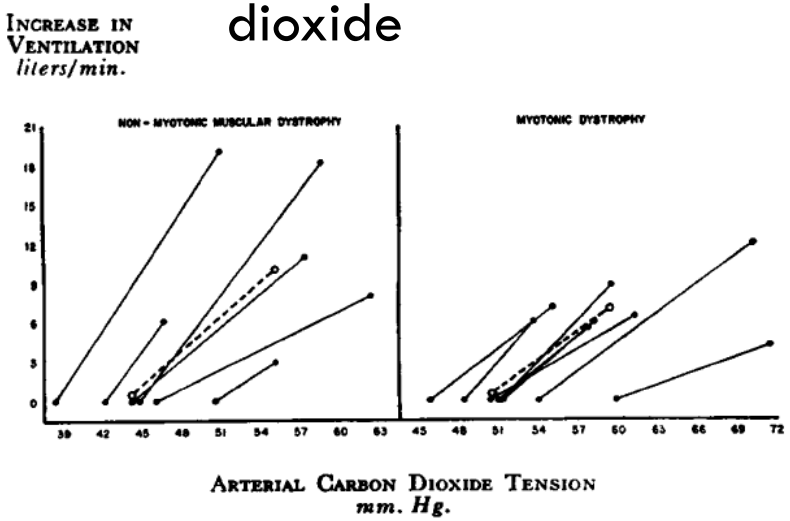

Perrin C, et al. Muscle Nerve 2004

# Respiratory Complications in MD

- Complications Includes
  - Pneumonia
  - Respiratory Failure (elevated CO2 or low oxygen)
  - Atelectasis (collapse of air sacks)

# Early Description of Pulmonary Complications:
## New England Journal of Medicine 1959

- 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

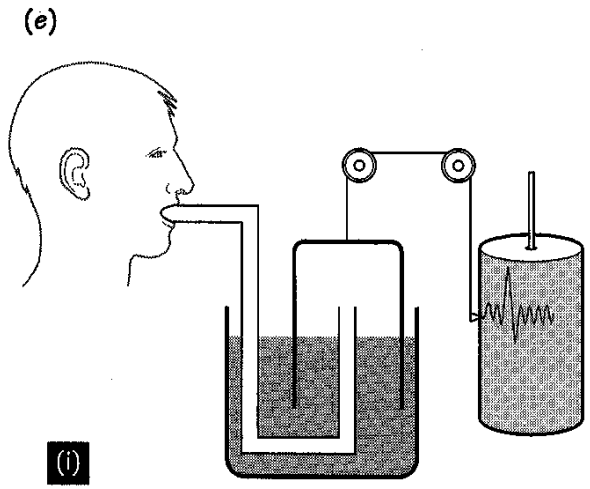
# What symptoms should I look for?

- 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)




water filled spirometer

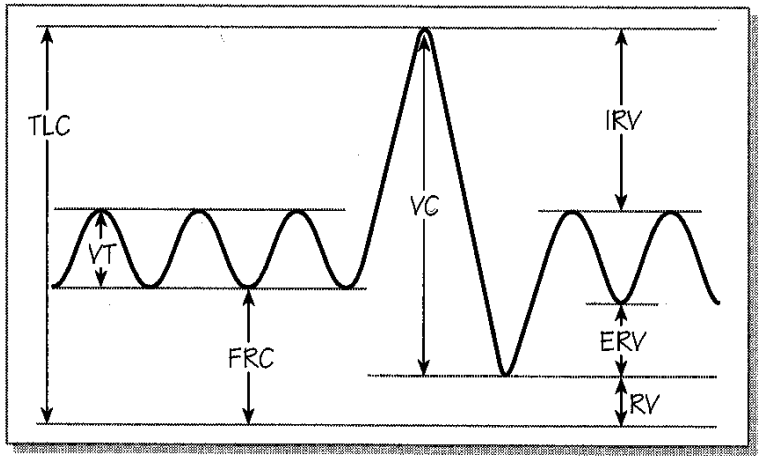



Table 1

| | | | |
|---|---|---|---|
| Tidal volume (VT) (at rest) | 500 mL | Total lung capacity (TLC) | 7300 mL |
| Vital capacity (VC) | 5500 mL | Functional residual capacity (FRC) | 3500 mL |
| Inspiratory veserve volume (IRV) | 3300 mL | Residual volume (RV) | 1800 mL |
| Expiratory reserve volume (ERV) | 1700 mL | | |

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

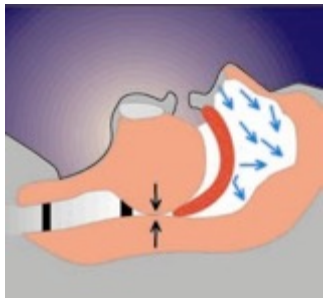
Obstructive Events

'Mixed'

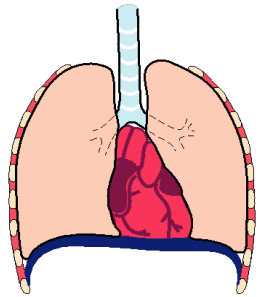
Central Events

'Cannot Breath'

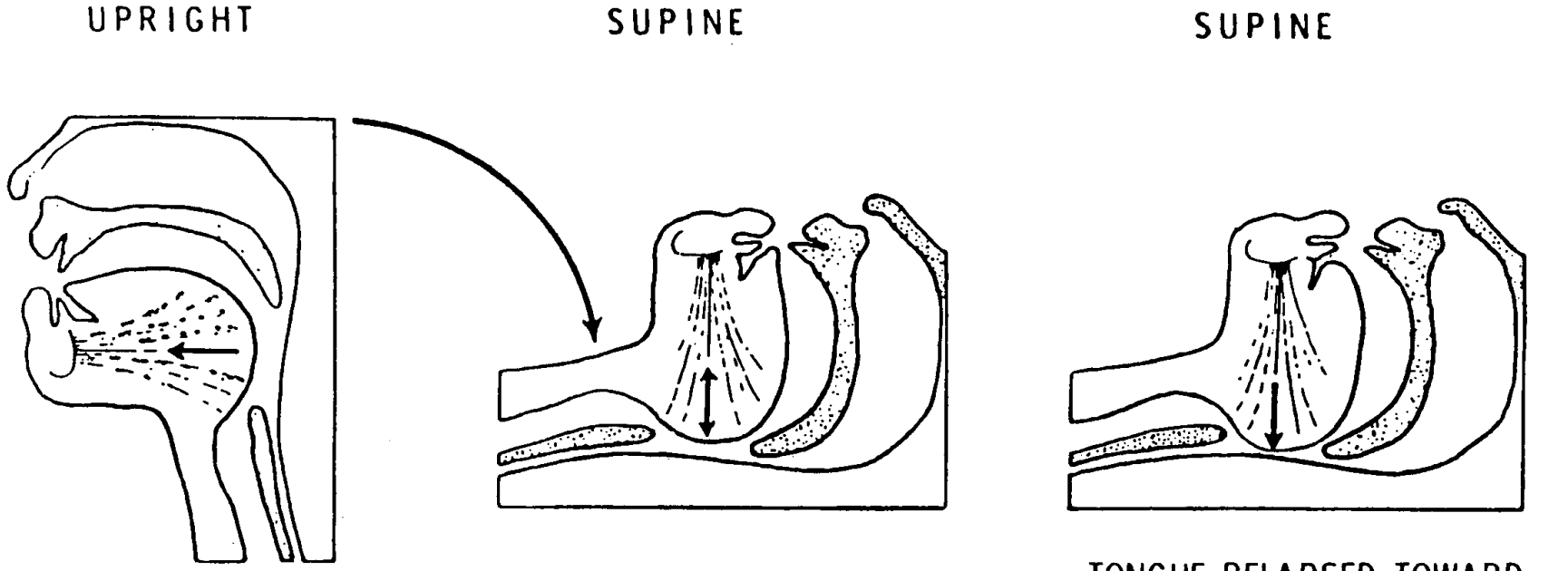
'Won't Breath'



Reduced Drive



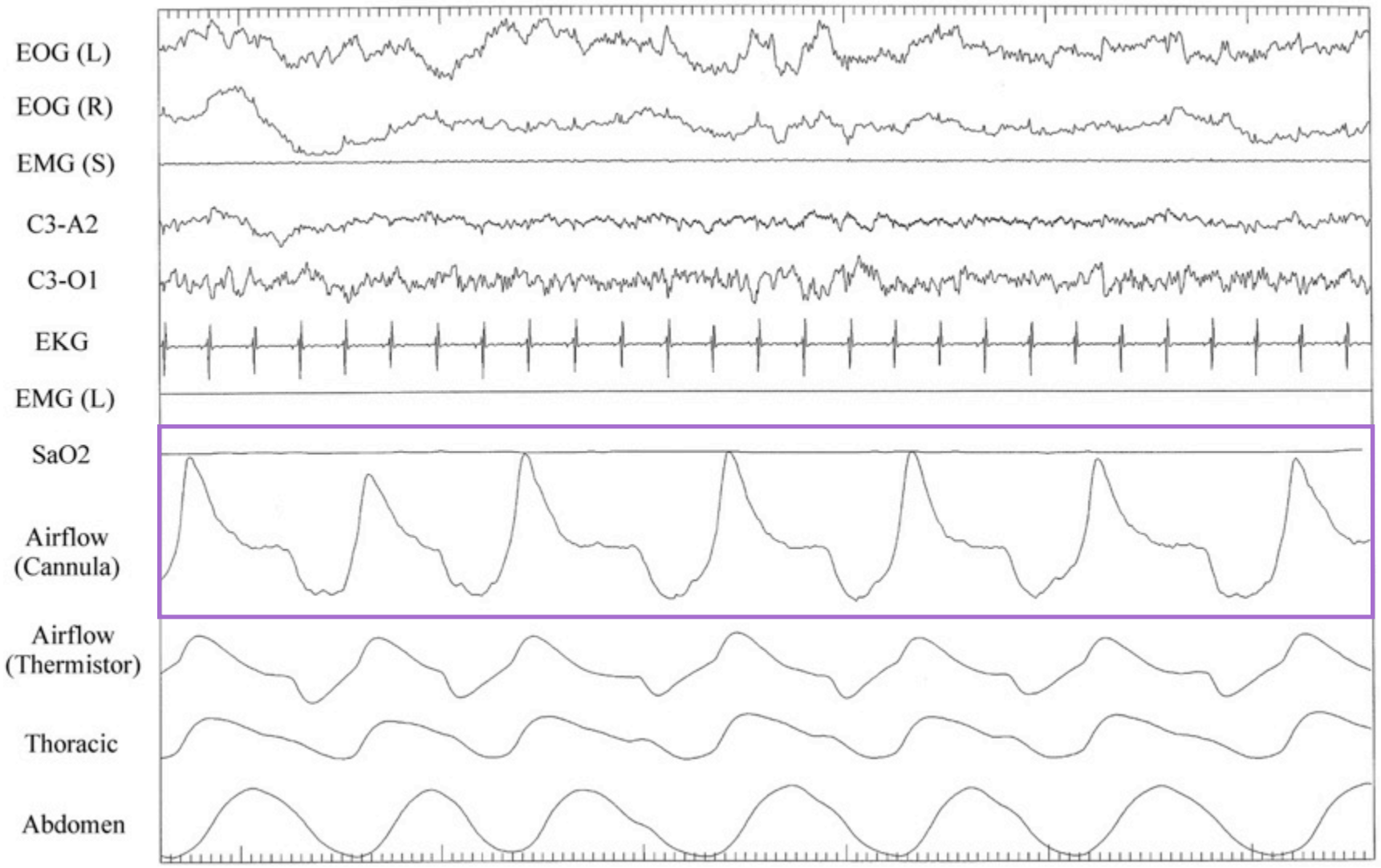
# Neuromuscular Activity and Upper Airway Obstruction



# 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



# 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