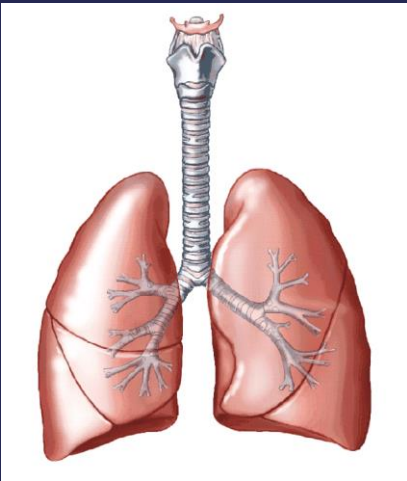


# KENNEDY DISEASE PULMONARY CONSIDERATIONS: SCIENCE & MANAGEMENT STRATEGIES



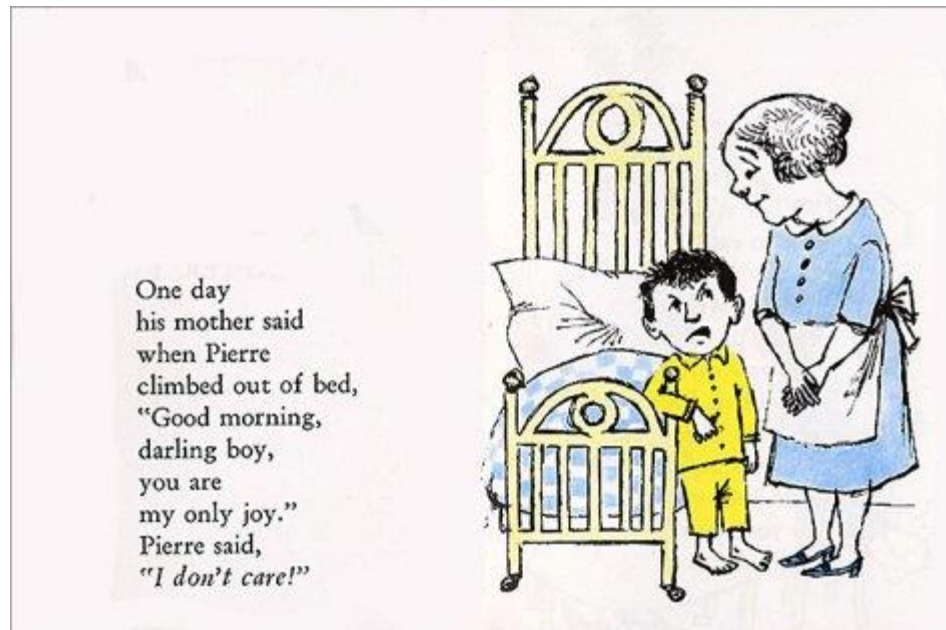
“When you can’t breathe nothing else matters”  
American Lung Association

# Overview

- Biology refresher course
  - ▣ The lungs and what they do
- What goes wrong in Kennedy Disease
- Evaluating breathing problems
- Evaluating sleep problems
- Treatment options
  - ▣ Lung recruitment
  - ▣ Ventilation
  - ▣ Secretion clearance
  - ▣ Sleepiness
  - ▣ Emergency plans

# Why Should I Care About This?

- Respiratory complications can cause functional impairment and lead death in Kennedy Disease



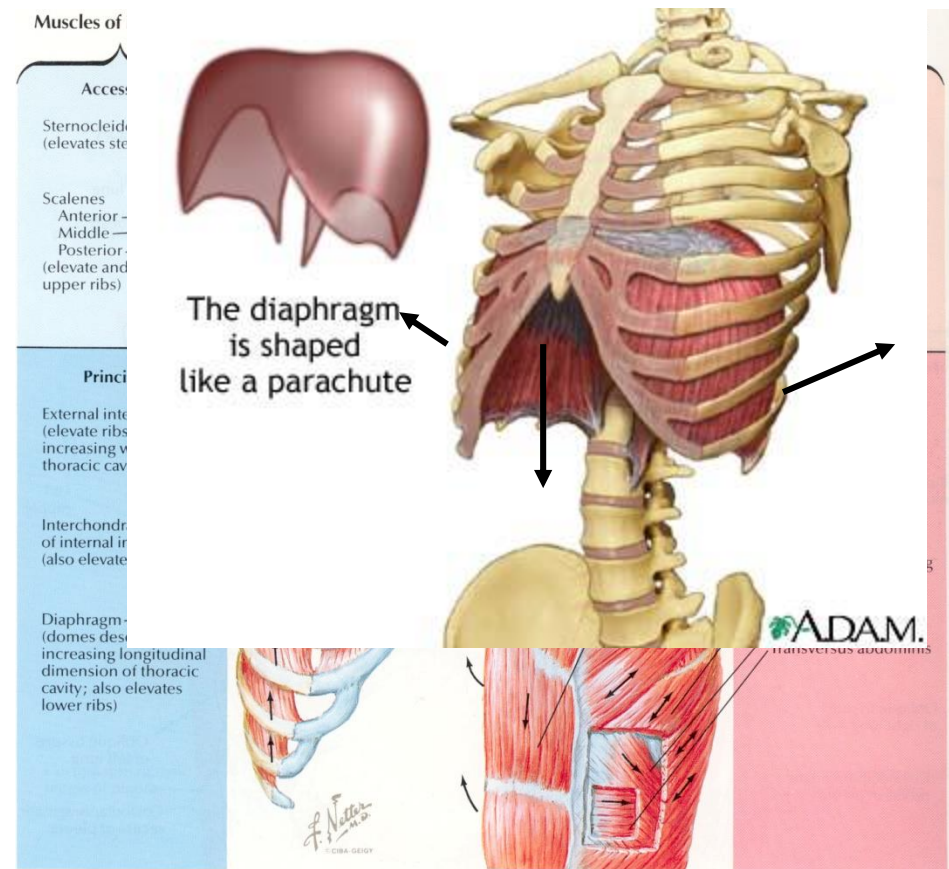
# Respiratory Muscles

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

- Diaphragm
- External intercostals
- Scalene, sternocleidomastoid

- **Expiratory:** Largely passive

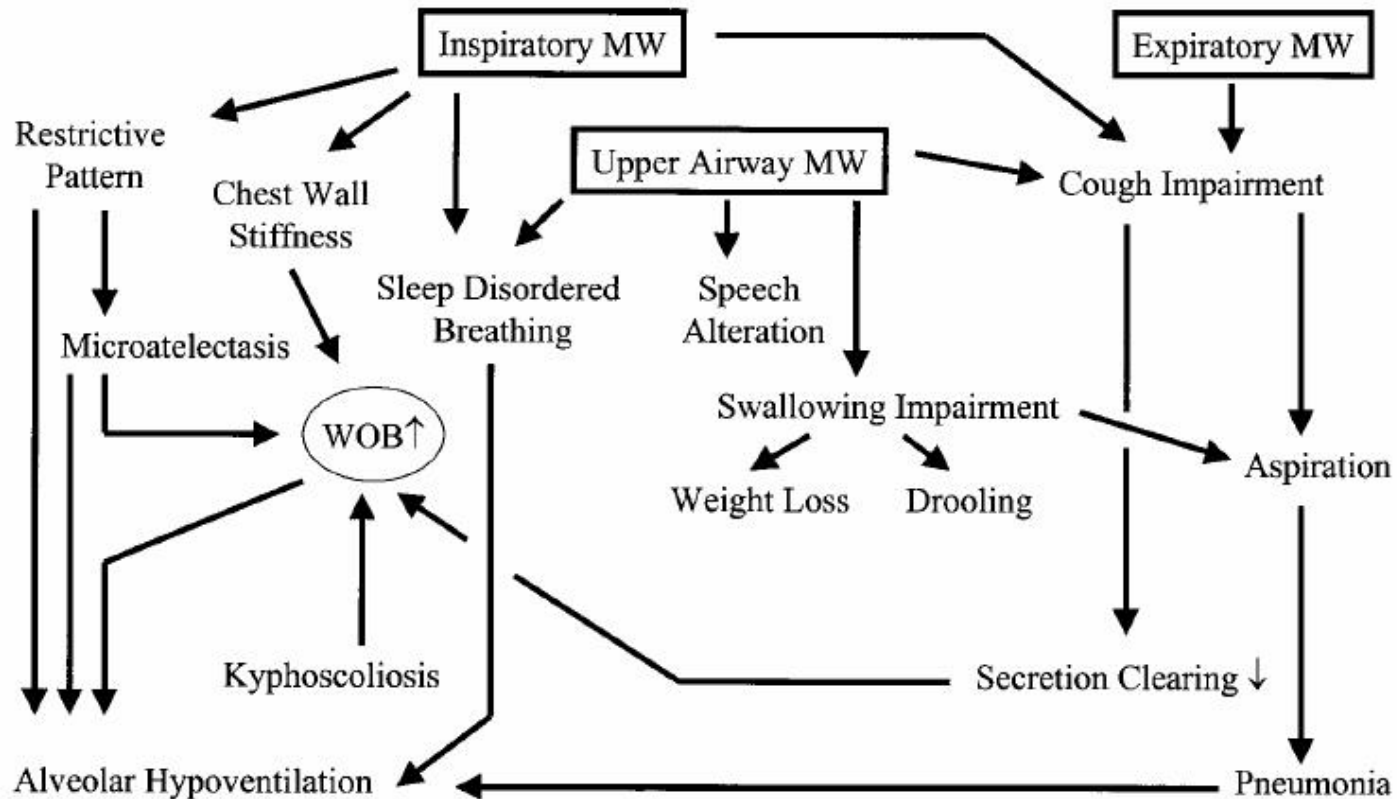
- Abdominal muscles
- Internal intercostals



# 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 (CO<sub>2</sub>)
  - ▣ When we exhale, we blow out carbon dioxide

# Respiratory Complications Are a Major Cause of Morbidity and Mortality in Neuromuscular Disorders



# Respiratory Complications in KD

- Complications Includes
  - Pneumonia
  - Respiratory Failure (elevated CO<sub>2</sub> or low oxygen)
  - Atelectasis (collapse of air sacks)

# Description of Natural Course of KD:

## Neurology 2008

- 39 patients at Mayo Clinic over 15 year period
- Survival at 10 years was 82%
- ~1/2 had shortness of breath
- 1/5 had orthopnea
- 1 of 25 used non-invasive ventilation
  - 0 used invasive ventilation



# Natural history of spinal and bulbar muscular atrophy (SBMA): a study of 223 Japanese patients

Atsuta, et al. Brain 2006

- Twenty One patients developed pneumonia
  - ▣ Mean age 62 years
- Fifteen died at mean age of 65

# General Considerations

- Respiratory complications can occur in KD
  - ▣ Can be due to respiratory muscle weakness
  - ▣ Can be due to bulbar weakness
- KD patients may not get routine pulmonary evaluation and testing
- Respiratory equipment is underutilized
- Sleep can be abnormal in KD
- Anesthesia can be problematic in KD

# 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)

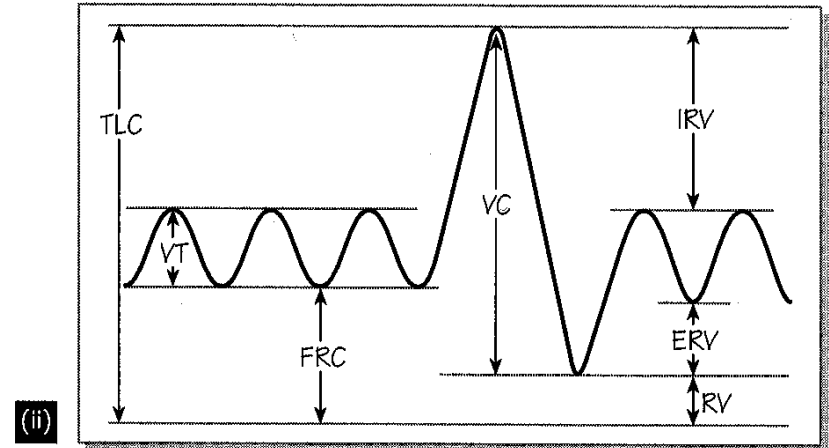
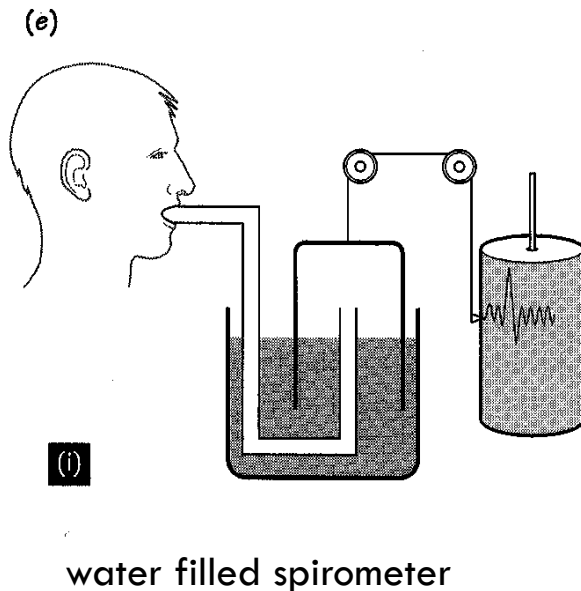


Table 1

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 reserve 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

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- Tests require a tight seal around a mouthpiece
- Weakness of facial muscles in KD can be a problem

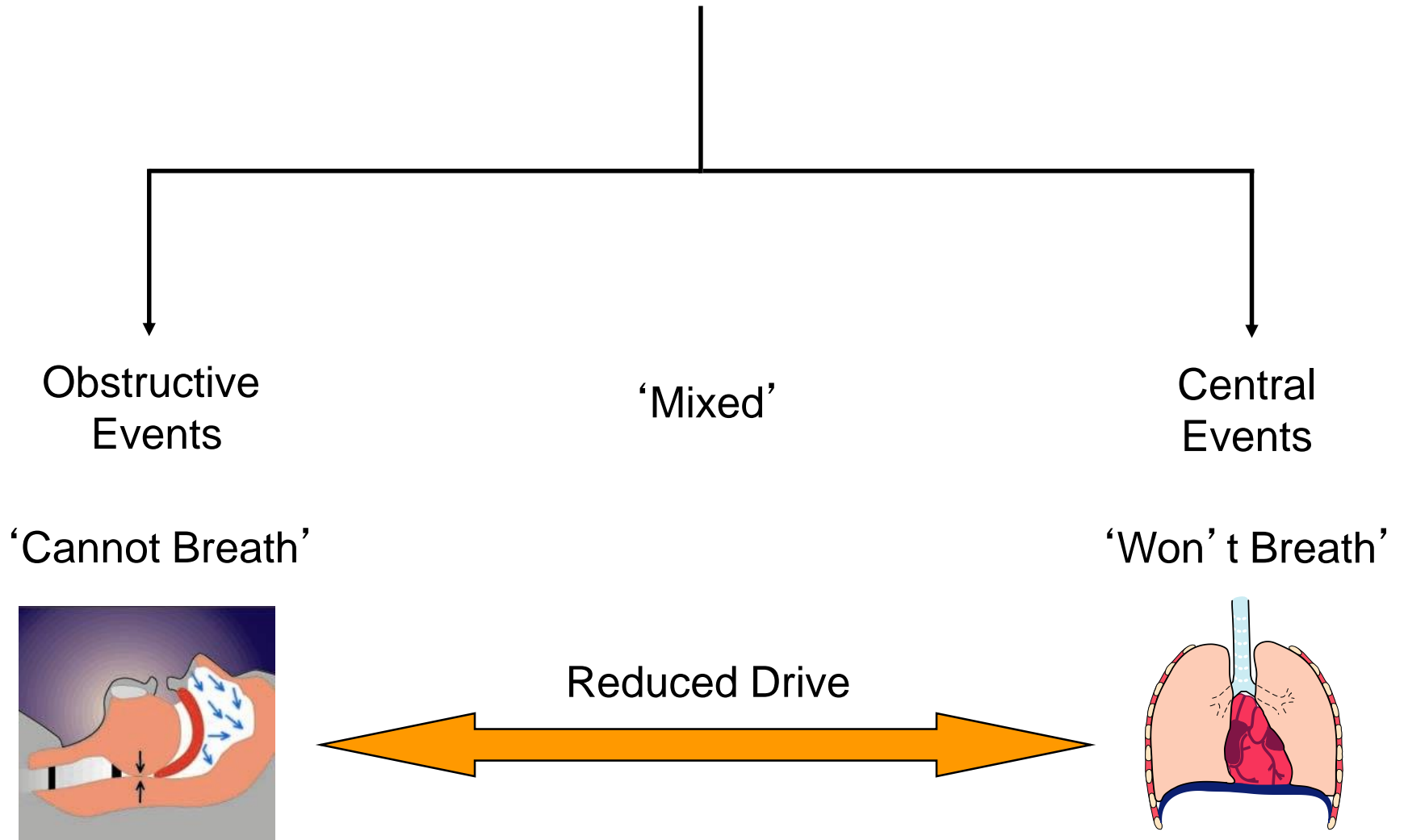


# Other pulmonary tests

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

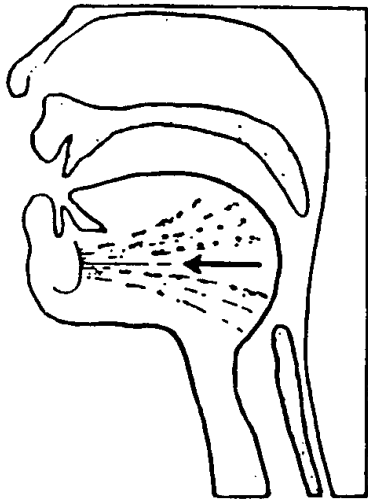
# Sleep Problems

# Terminology of Sleep Apnea



# Neuromuscular Activity and Upper Airway Obstruction

UPRIGHT

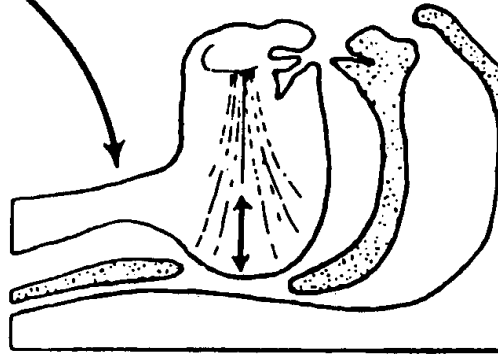


AIRWAY OPEN



Genioglossal EMG:  
INSPIRATORY BURSTS

SUPINE

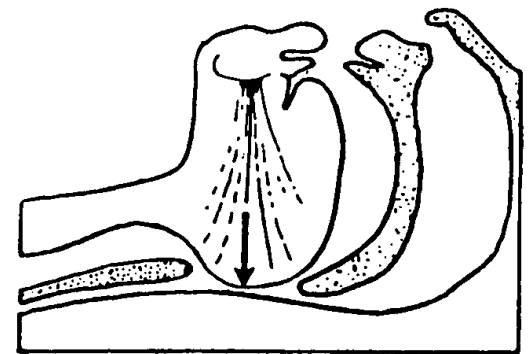


TENDENCY OF TONGUE TO RELAPSE  
AIRWAY OPEN

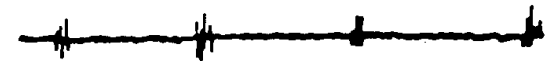


Genioglossal EMG:  
TONIC ACTIVITY  
+ INSPIRATORY BURSTS

SUPINE



TONGUE RELAPSED TOWARD  
POSTERIOR PHARYNGEAL WALL  
INCREASED AIRWAY RESISTANCE



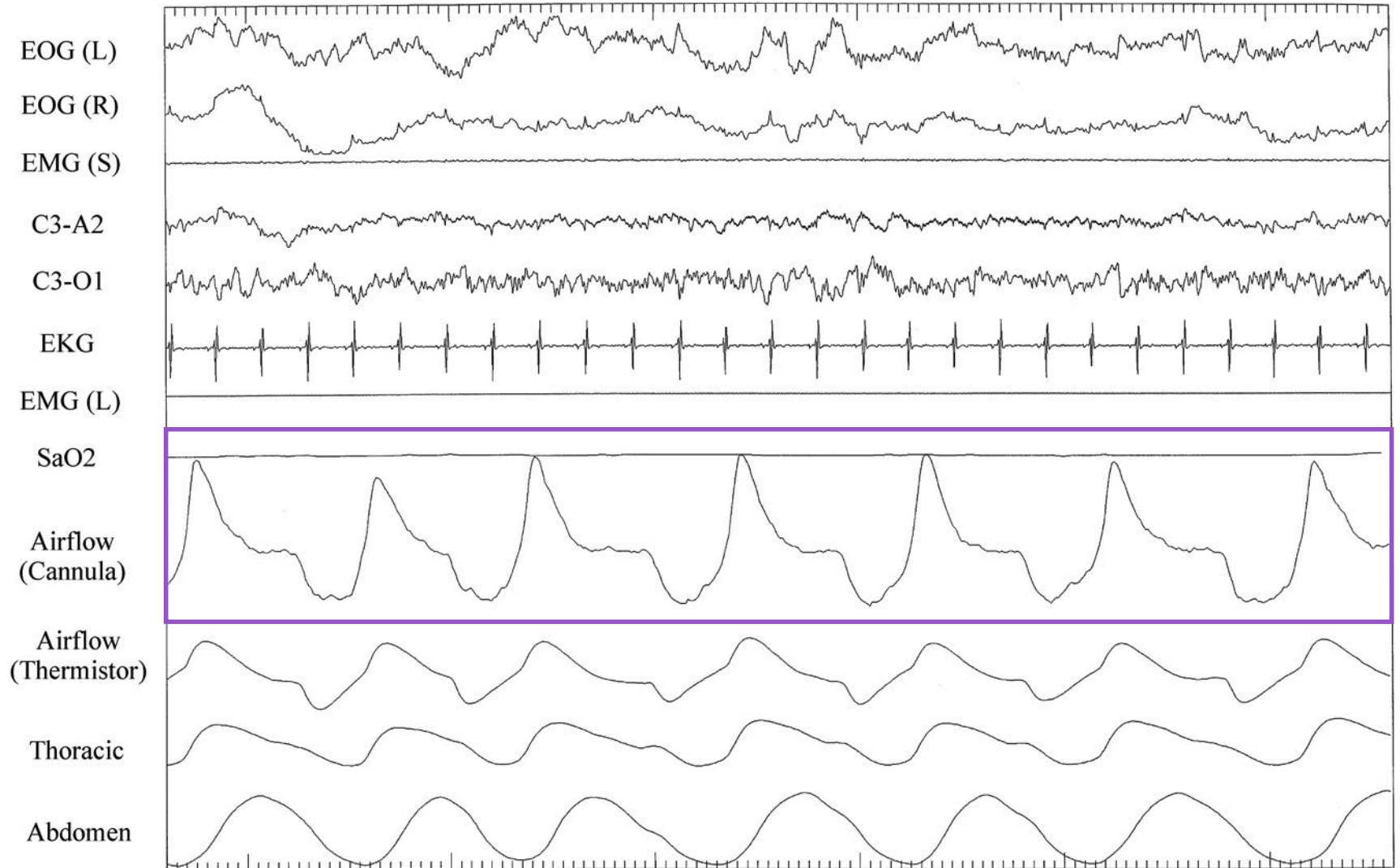
Genioglossal EMG:  
GREATLY DIMINISHED OR  
ABSENT ACTIVITY

# 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



# 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)



# 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 Kennedy Disease

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- 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, PaCO<sub>2</sub>>45
  - May have benefit at earlier times
  - May be better to target symptoms

# Surgery

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


# Prevention

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

# Emergency Preparedness



- [www.ventusers.org](http://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 KD than providers
  - Need to be an advocate

# Case 1: J.H. - 80yo man

- I met in 2005, initially diagnosed with myasthenia gravis in 1998
- Initial symptoms were eye droop
- Developed voice changes and shortness of breath
- Dx'd with Kennedy's in 2006
- Had dx of sleep apnea but didn't tolerate CPAP
- Normal lung function in 2008, but moderately reduced in 2014
- CO2 levels persistently elevated
- Started BiPAP and he sleeps better and has less SOB

# Case 2: P.N. 62 yo man

- Diagnosed at age 59 but symptoms began in his 20s
- Has limb weakness and shortness of breath with exertion
- Has sleep apnea and is using CPAP
- Has mildly reduced lung function
- No pulmonary interventions yet

# Summary

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- Breathing involvement in KD is highly variable
- Weakness of inspiratory and expiratory muscles can occur
- Illness and death due to breathing complications occurs
- Routine pulmonary evaluation is important
- Treatment may include CPAP, BiPAP, assisted cough
- Sleep evaluation is also important



# Questions

# Control of Ventilation (Breathing)

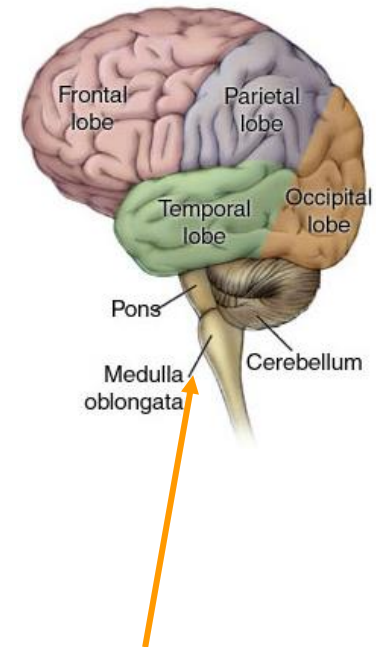
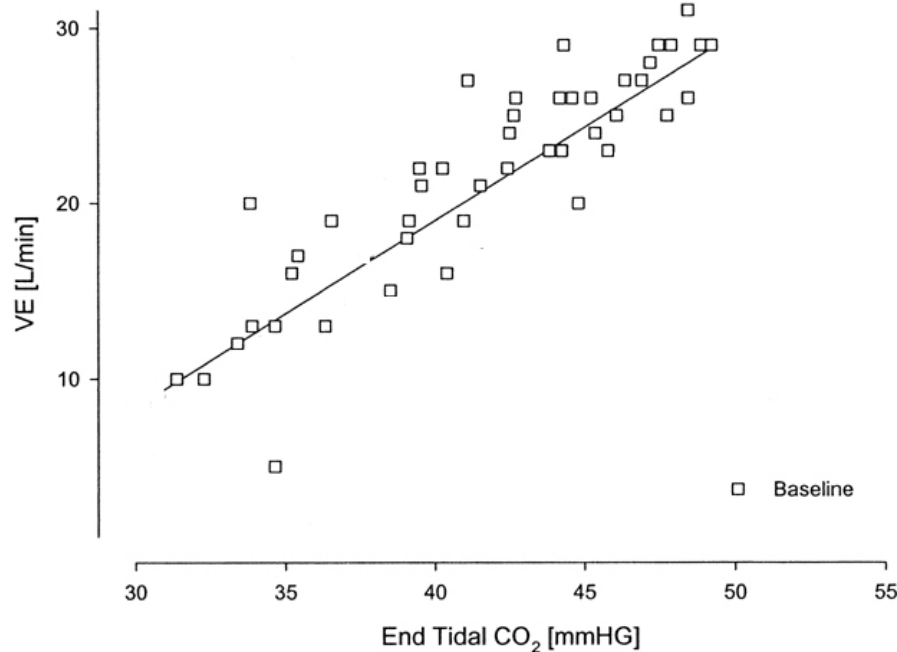
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# Control of Ventilation (Breathing)

## □ 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 KD 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 KD should not receive oxygen without ventilatory support or close monitoring