Respiratory Management of Facioscapulohumeral Muscular Dystrophy

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Respiratory Involvement in FSHD

- Very variable
  - time of onset
  - rate of progression
  - Muscles involved – may be assymetrical
  - Probably related to epigenetics
- Sleep apnea a common manifestation
- Respiratory failure seen in a small %
- Need for mechanical ventilation only 1% in a Dutch study

Respiratory Muscles affected by FSHD

- Facial Muscles
- Bulbar muscles
  - Speech and swallowing
- Inspiratory muscles
  - Diaphragm, chest and neck
- Expiratory muscles
  - Abdominal and chest

Upper Airway

Bulbar Muscles; Speech Swallowing

Epiglottis

Vocal Cords

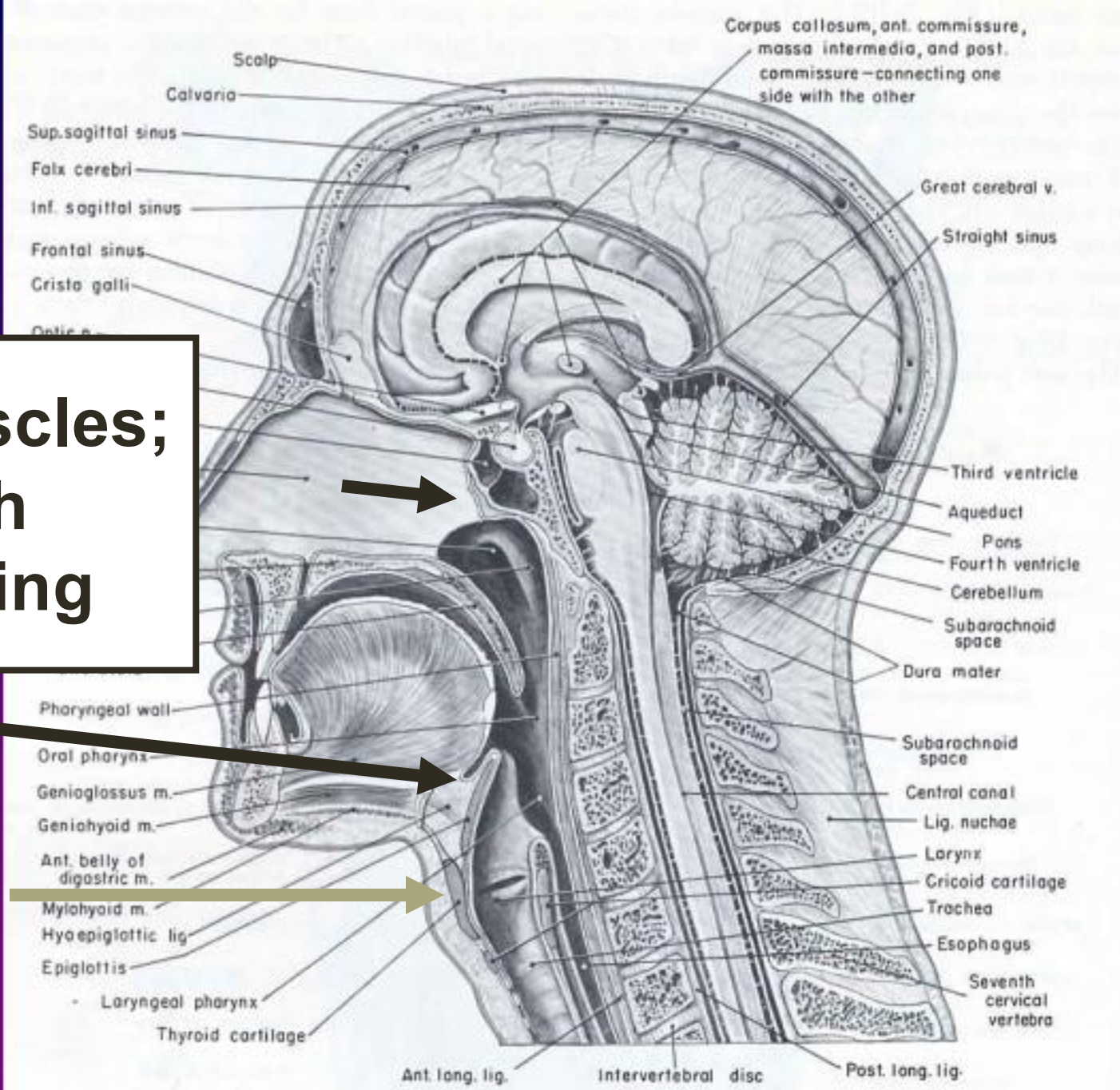
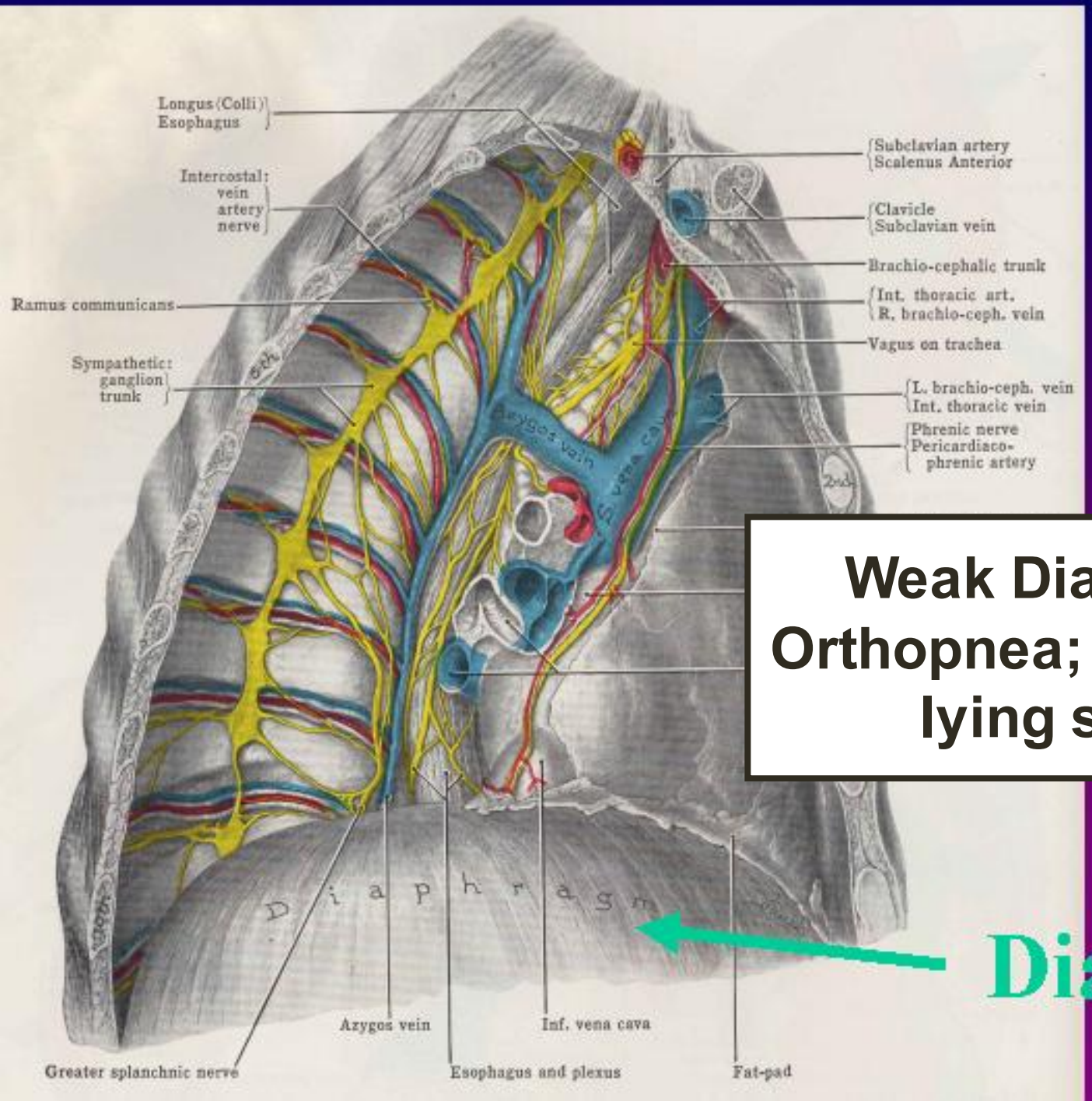


Fig. 7–108. Sagittal section of the head and neck showing the visceral tube.



Weak Diaphragm: Orthopnea; SOB when lying supine

Diaphragm

Accessory Muscles: Inspiratory Muscles used when the diaphragm becomes stressed or weakened

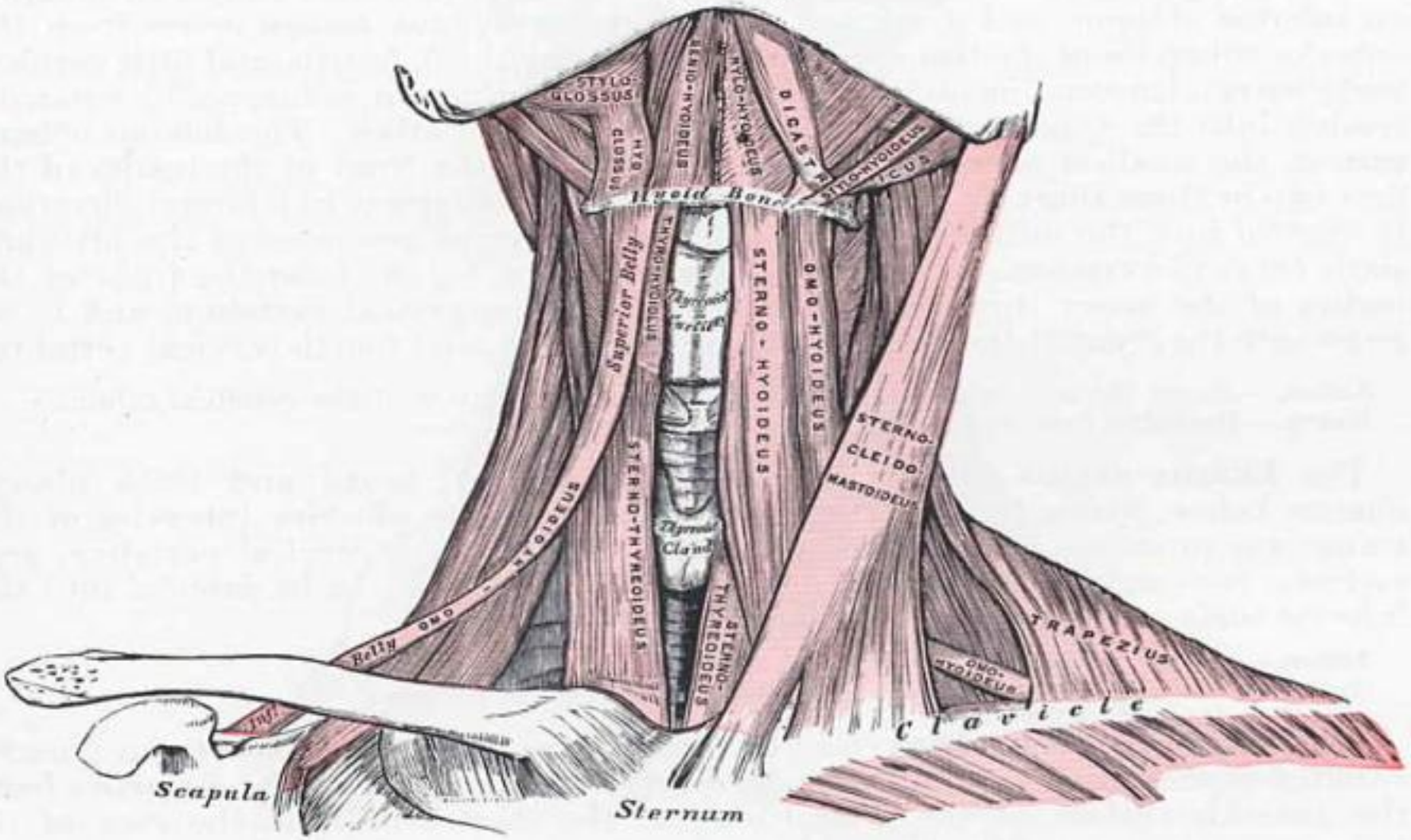
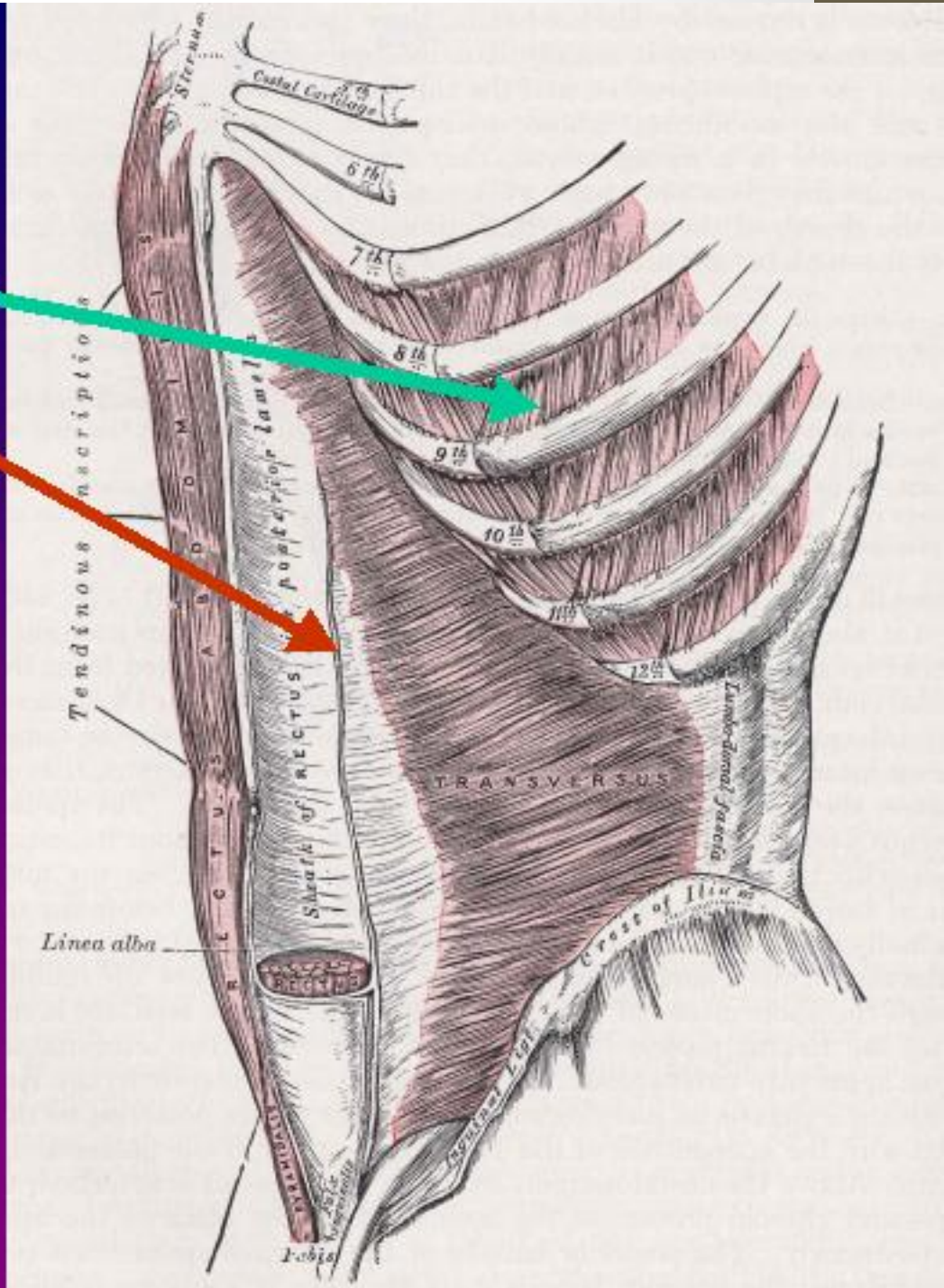


FIG. 434.—Muscles of the neck. Anterior view.

Expiratory Muscles
Intercostals
Abdominals

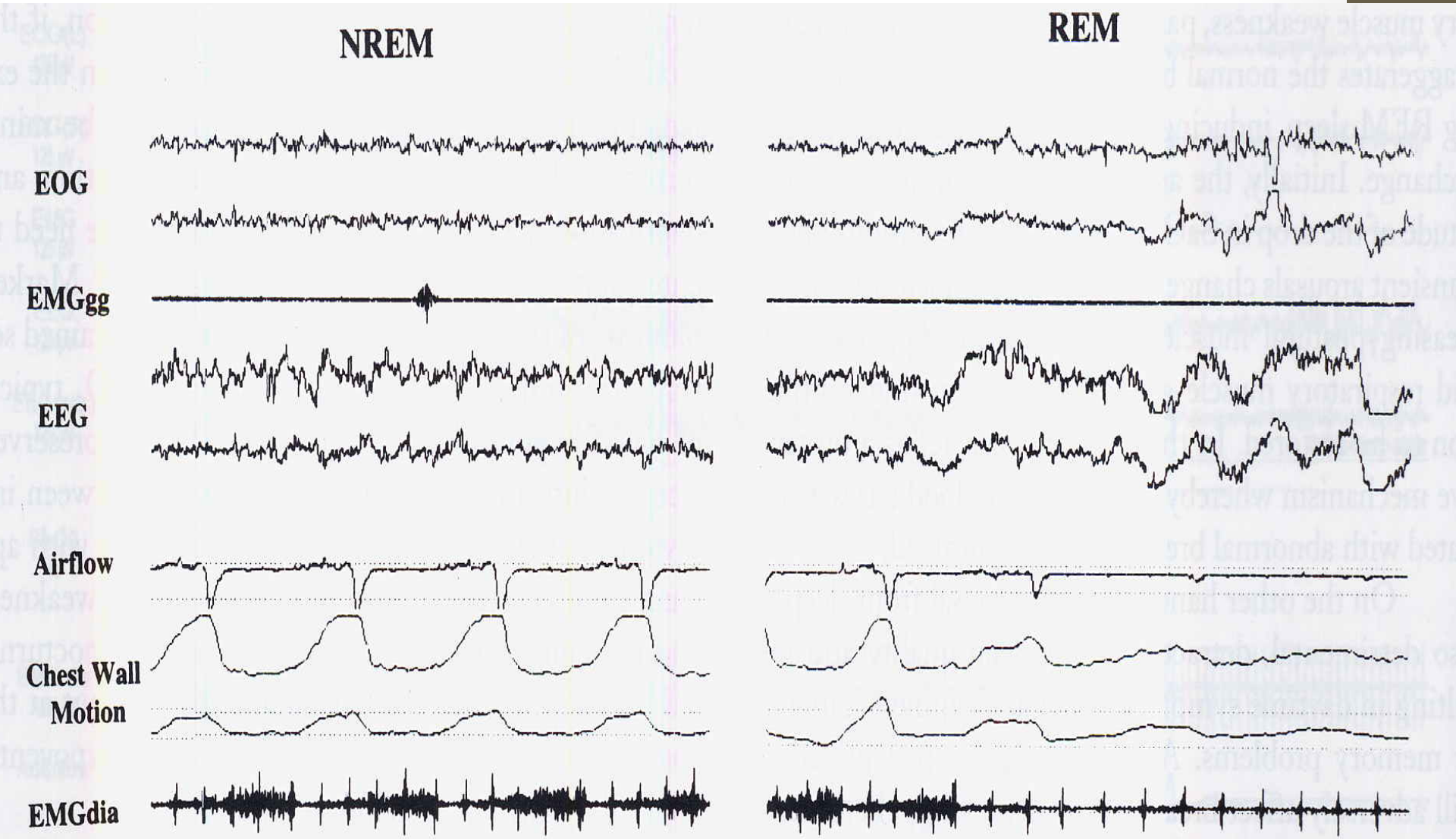
Also need for effective Cough:
2) inspiratory muscles
3) adequate bulbar function



Effects of FSHD on Sleep

- Normal sleep – reduces drive to breathe, muscle tone down including upper airways, CO2 increases
- More slowing of breathing when muscles are weakened
- Upper airway more prone to closure
- Sleep apnea a risk
- During rapid eye movement (REM) sleep, other breathing muscles become flaccid and if diaphragm is weak or paralyzed, breathing ceases – patients become REM deprived
- Excessive weakness of muscles leads to excessive CO2 retention at night, then day and night

Effect of REM in NMD pt with Diaphragm Dysfunction



Typical Symptoms of Respiratory Muscle Involvement by FSH

- Shortness of breath
  - Exertional, at rest, or positional
- Poor sleep
  - Snoring, morning headaches
  - Fatigue, sleepiness
- Swallowing Problems
  - Choking, aspiration
  - Wheezing or stridor
- Weak cough

Evaluation of Patients with FSH and Respiratory Involvement

- **Medical exam**
  - History
  - Physical findings (muscles, cough)
- **Pulmonary Function Tests**
  - Spirogram (Vital Capacity)
  - Insp and exp pressures
  - Lung volumes
- **Gas exchange**
  - Oximetry: % of hemoglobin carrying oxygen
  - Blood Gases: Carbon dioxide tension ($PaCO_2$), Oxygen tension (PaO_2) (nl values)

Evaluation of Patients with FSH and Respiratory Symptoms

- Sleep Study
  - NocturnalOximetry
  - Polysomnogram/ home studies
- Upper Airway evaluation
  - Modified Ba swallow
  - Laryngoscopy
- Diaphragm Function
  - Upright and supine PFTs (Face Mask)
  - Diaphragmatic Pressures
  - Sniff test

Therapy of FSHD – Respiratory Involvement

- Preventive
  - Vaccines – flu yearly/ pneumovax 5 yrs
  - Careful swallowing, assist cough
  - Nutrition – Speech and swallow eval
  - ? Role for albuterol
  - ? Gene therapy

Facial, Bulbar Muscle Weakness

- Impairs speech and swallowing
- Treatment:
  - Swallowing therapy
    - Thickeners, soft mechanical food
    - Chin tuck

Therapy of FSHD – Respiratory Involvement

- Sleep Apnea
  - Continuous Positive Airway Pressure (CPAP)
  - Mandibular advancement
  - Oxygen therapy (with caution)

PRESSURE APPLIED THROUGH THE NARES

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THE LANCET, APRIL 18, 1981

Original Report of CPAP to treat Obstr Sleep Apnea

Therapy of FSHD – Respiratory Muscle Involvement

- **Inspiratory muscle weakness: Respiratory Failure**
  - **Night only or day as well**
  - **Increased $PaCO_2$**
  - **Decreased PaO_2**

- **Therapy**
  - **Noninvasive Ventilation (BiPAP)**
  - **Invasive Mechanical Ventilation**

Therapy of ALS – Noninvasive Ventilation ("BiPAP™")

- "Interface"
  - Nasal Mask
  - Full face mask
  - Mouthpiece
- Ventilator
  - Small, portable "bilevel"
  - Volume ventilators

Interfaces for CPAP/NIV

Ventilators for NIV

Expiratory Muscle Weakness

- Impairs cough effectiveness
- Treatment:
  - Manually-assisted coughing
  - Cough Assist (inexsufflator)
- Lethal if combined with severe bulbar involvement

Cough Assist

T70

Initiation of NIV: suggestions

- Comfortable mask
- Ventilator; compact, quiet, portable
- Pressures low, gradually work up
- Start during daytime,1 or 2 hrs until feels OK
- Initiate nocturnal use gradually
- Routine humidification
- Treat it like a musical instrument
- May take weeks or months

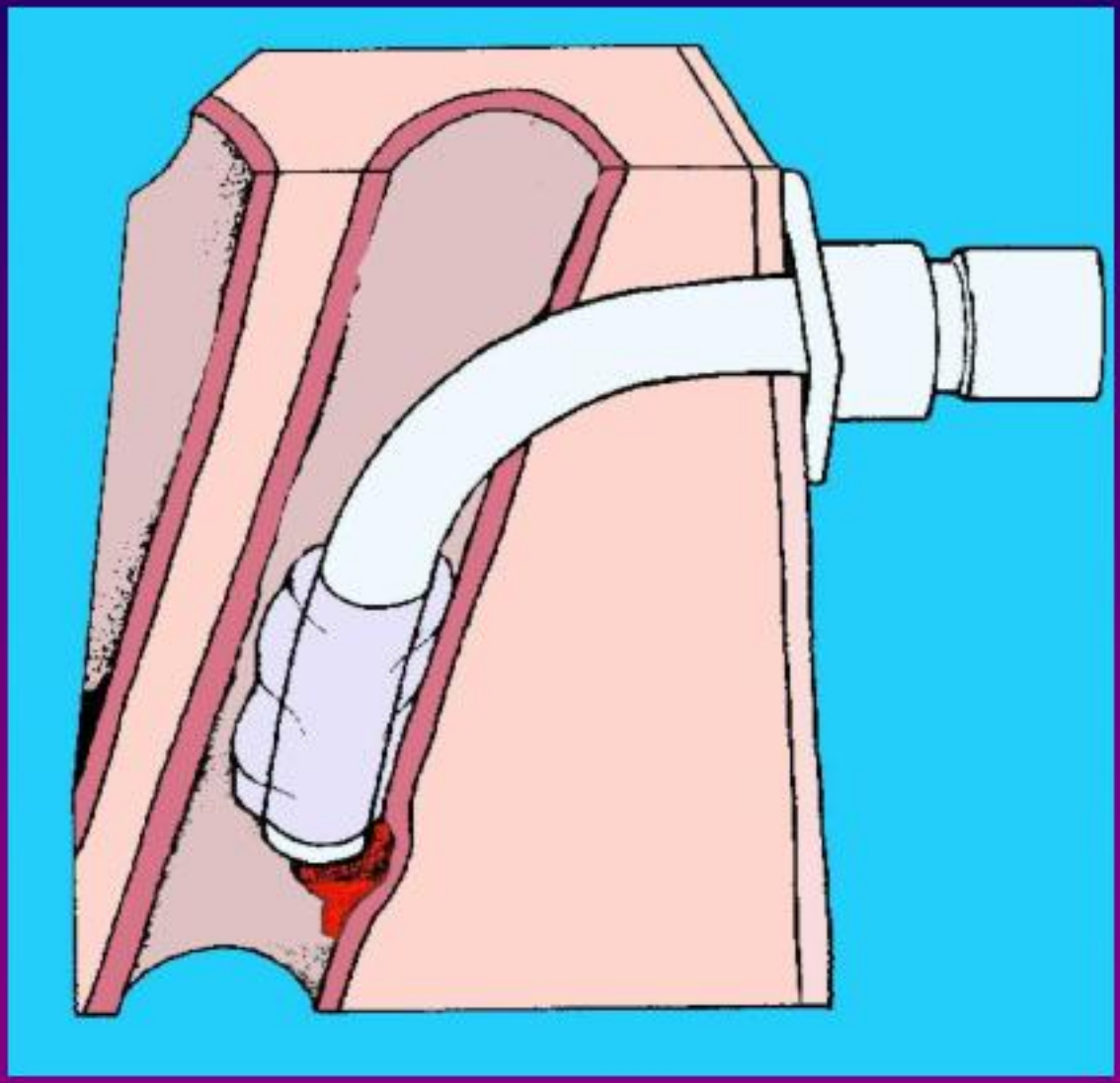
Therapy of FSH – Invasive Ventilation

- For patients who desire it and can't tolerate, failed or have contraindications to NIV:
  - Tracheostomy tube: Plastic or metal tube placed surgically in the windpipe
  - Requires maintenance, suctioning, must be changed every 4-8 wks
  - Cuff may cause tracheal damage
  - Increases risk of infection
  - More challenging to manage for caregivers

Standard Trache Tubes: Portex Single Channel Cannulae, Shiley - inner cannula

Speaking Valves

Tracheal Trauma from Mis-shapen Tubes



Summary

- Quite variable in onset, presentation
  - Facial/Bulbar muscles
  - Inspiratory muscles/Diaphragm
  - Expiratory muscles
  - Ventilatory support 1%

- Therapy includes:
  - CPAP or other approaches for sleep apnea
  - NIV first rx for insp muscle weakness
  - Invasive mechanical ventilation is used less
  - Expiratory muscle weakness impairs cough, cough assistance can help
  - ?albuterol; Future: Gene therapy