



Respiratory Care of Children with Rare Diseases

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No disclosures or COIs

Objectives

- Swallowing dysfunction, aspiration and recurrent respiratory symptoms
- Impaired cough and mucociliary clearance
- Sleep disordered breathing associated with airway abnormalities, hypotonia, obesity and control of breathing abnormalities
- Neuromuscular disease and restrictive lung disease

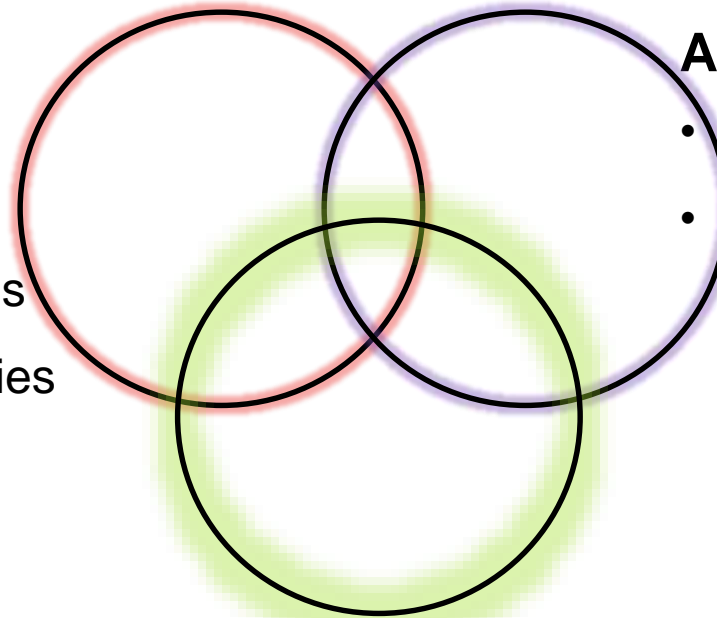
Understanding respiratory morbidity in children with Mobius syndrome

- Mobius syndrome is a rare disease
- No prospective studies have been done on natural history of lung function and lung decline in people with Mobius syndrome
- Great variability exists regarding respiratory symptoms in children with Mobius syndrome exist
- Lessons learned from other diseases with similar respiratory co-morbidities

Factors that may influence respiratory disease and severity

Host Characteristics

- Co-morbidities
- Swallowing dysfunction
- Neuromuscular weakness
- Upper airway abnormalities



Age/Timing of Exposure

- Growth and Development
- Susceptibility to injury

Environmental and Social Factors

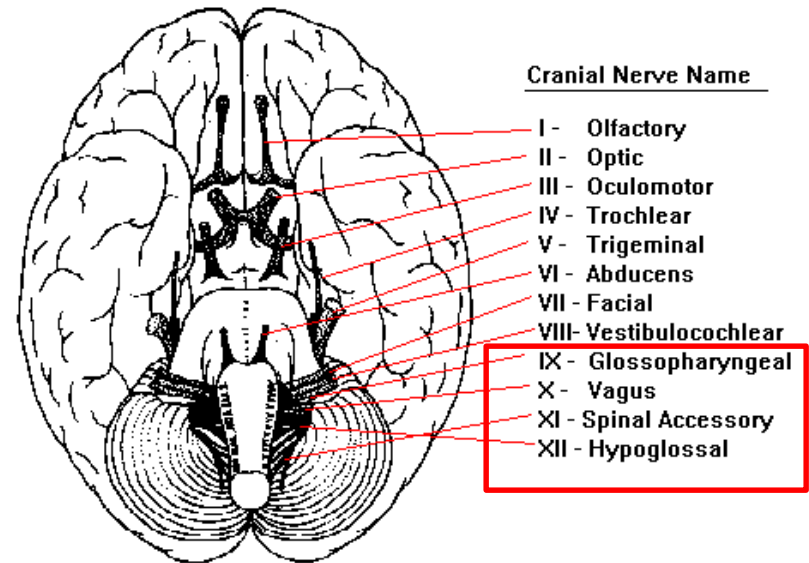
- Viral illnesses/ exposure to secondhand smoke
- Health care access and management
- Exposure to other environmental stressors

Respiratory symptoms associated with chronic diseases of childhood

- Impaired swallowing (dysphagia) and **recurrent aspiration**
- **Poor cough and clearance of airway secretions**
- Poor nutrition and **prolonged respiratory symptoms with viral infections**
- **Upper airway obstruction, neuromuscular weakness, hypotonia**

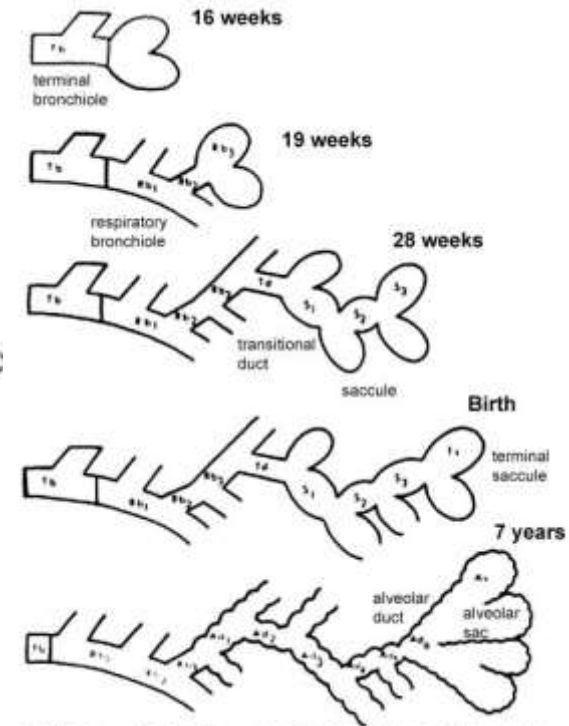
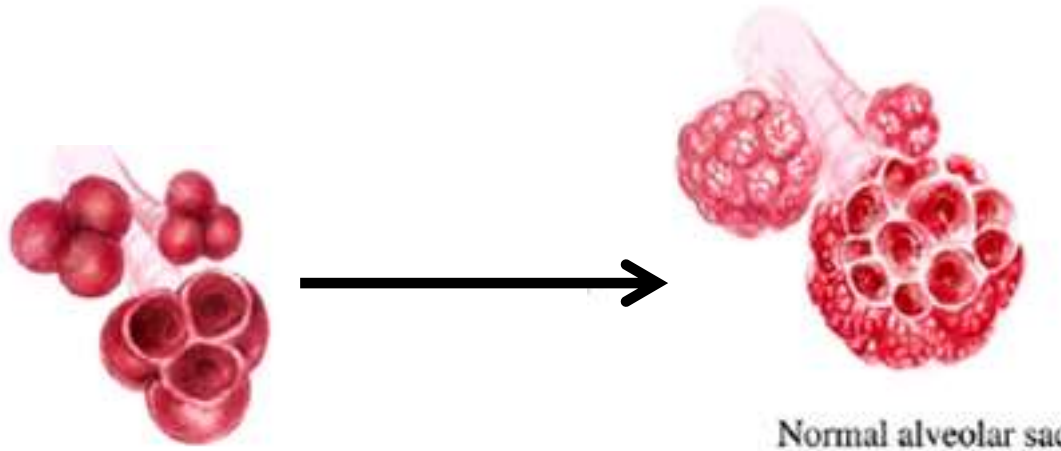
Extent of cranial nerve involvement can influence respiratory symptoms

- Impairment of cranial nerves, 9,10,11 and 12 in addition to 6 and 7
 - Swallowing and chewing difficulties
 - Tongue movement abnormalities may increase **risk of aspiration** and cause failure to thrive
 - Nasal regurgitation
 - Trouble swallowing liquids
 - Dysphonia and dysarthria
 - Impaired gag reflex



The developing lung and aspiration

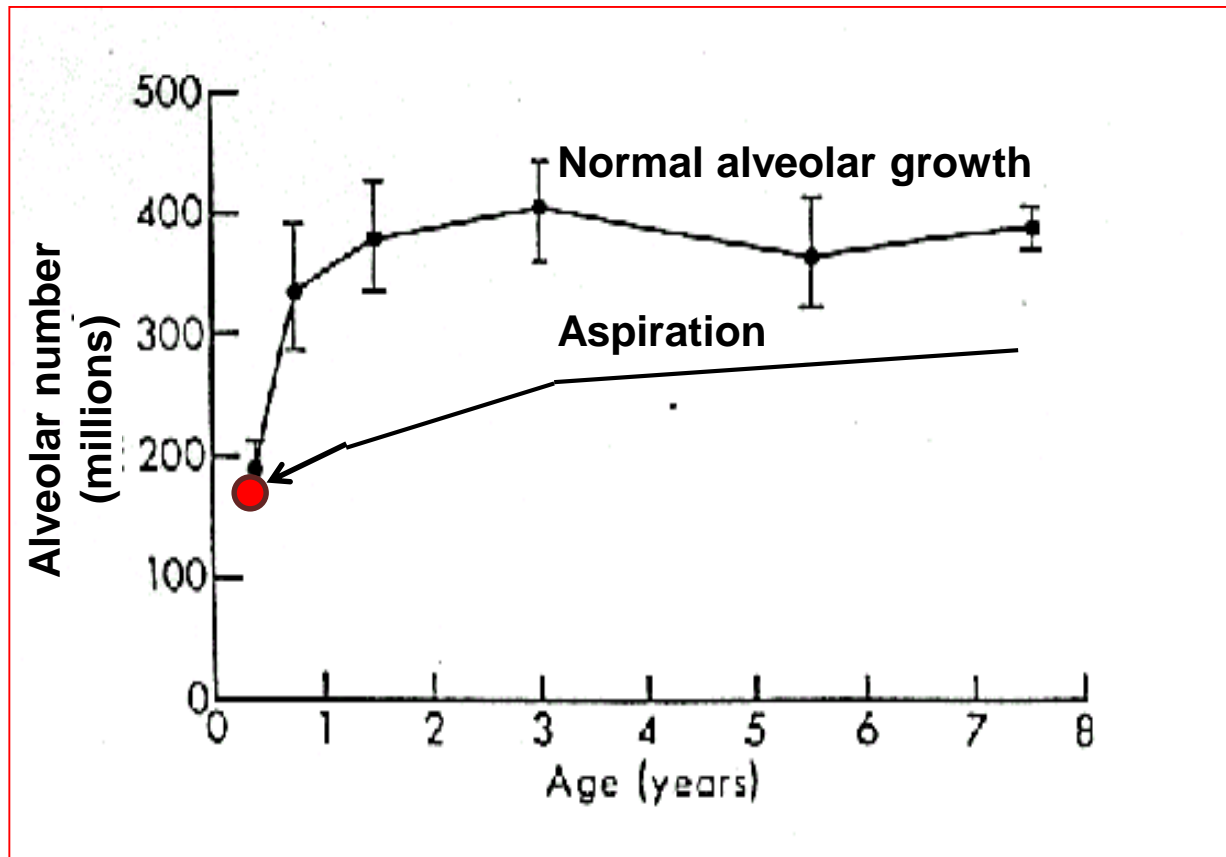
Recurrent aspiration may impair alveolar growth in infants/children



Modified from Dilly SA. Thorax. 1984 Oct;39(10):733-42. PMID: 6495241

Adapted from Thebaud and Abman, AJRCCM, 2007
http://embryology.med.unsw.edu.au/embryology/index.php?title=Lecture_-_Respiratory_Development

The majority of postnatal alveolar growth occurs during the first two years of life



Adapted from Thurlbeck W.M. Thorax 37:564-571, 1982

Symptoms of dysphagia and aspiration in infants and young children

- Chronic wheezing and/or cough that does not or only partially responds to medications
- Failure of disease resolution
 - Chronic chest xray changes that persist including:
 - Interstitial changes, atelectasis or hyperinflation
- Poor weight gain, including food refusal

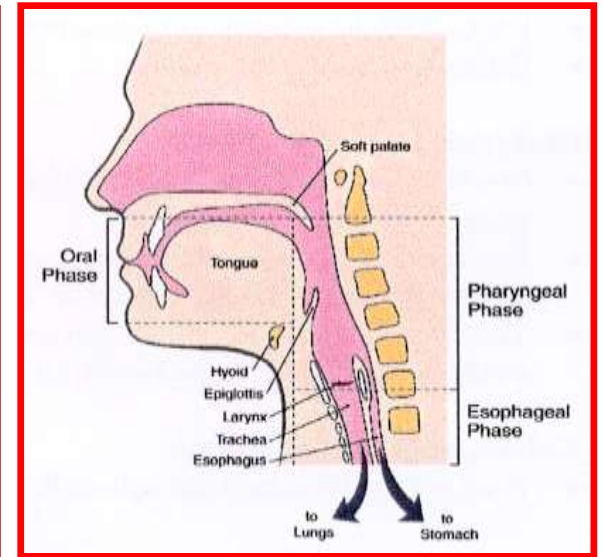
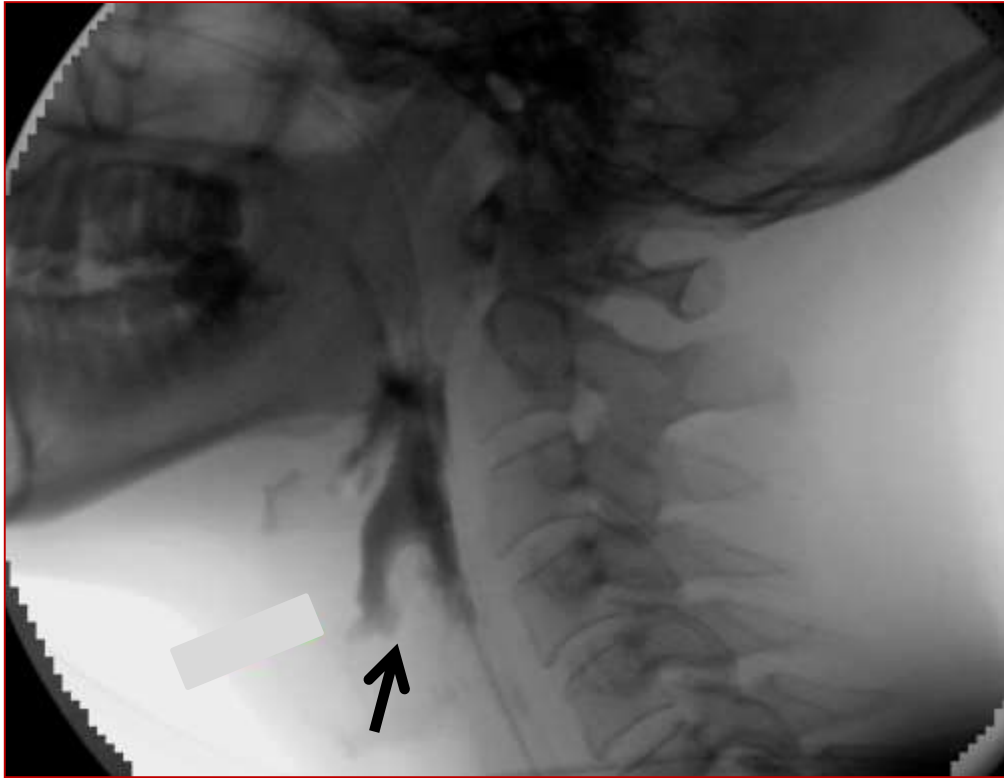
Symptoms of dysphagia and aspiration in infants and young children

- Oxygen desaturation with feeds, exertion and sleep
- Coughing and tachypnea (fast breathing) with feeds
- Increased severity of respiratory symptoms with common respiratory viruses- such as RSV or rhinovirus
 - Increased respiratory rate and work of breathing may exacerbate swallowing dysfunction or aspiration

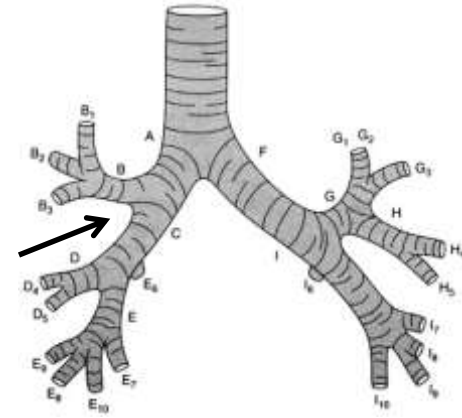
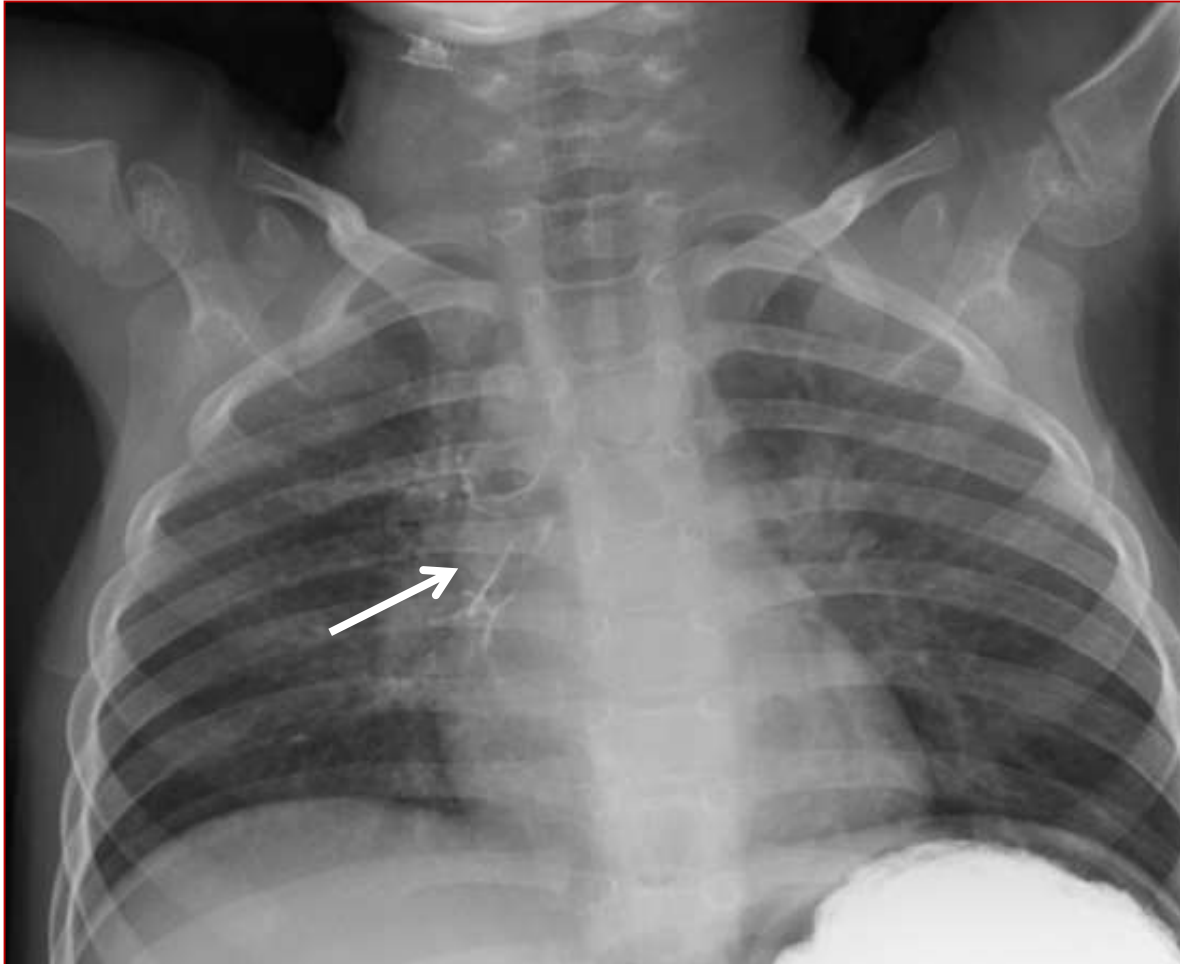
Common tests/strategies used to detect dysphagia with aspiration in infants/children

- * Videofluoroscopic swallow study
- * Flexible endoscopic evaluation of swallowing (FEES)
- * Bronchoscopy
 - Fat-laden macrophages and airway inflammation- not specific
- * Clinical response
 - Enteral feeds to assess clinical response
 - Transpyloric feeds, continuous GT feeds

Laryngeal penetration increases risk of aspiration



Aspiration into right upper lobe and bronchus intermedius



Other factors that may influence severity of respiratory symptoms in children with aspiration

- Gastroesophageal reflux
- Airway abnormalities- interfere with normal function
 - Undetected tracheal clefts
 - Vocal fold abnormalities
 - Impaired mucociliary clearance and chronic infection
- Neurological impairment
 - Abnormal tone and inability to coordinate swallowing
 - Impaired protective airway responses- such as silent aspiration

Other factors that can influence severity of respiratory symptoms in children with aspiration

– Age of swallowing dysfunction and duration

- May have greater impact on lungs during the first two years of life

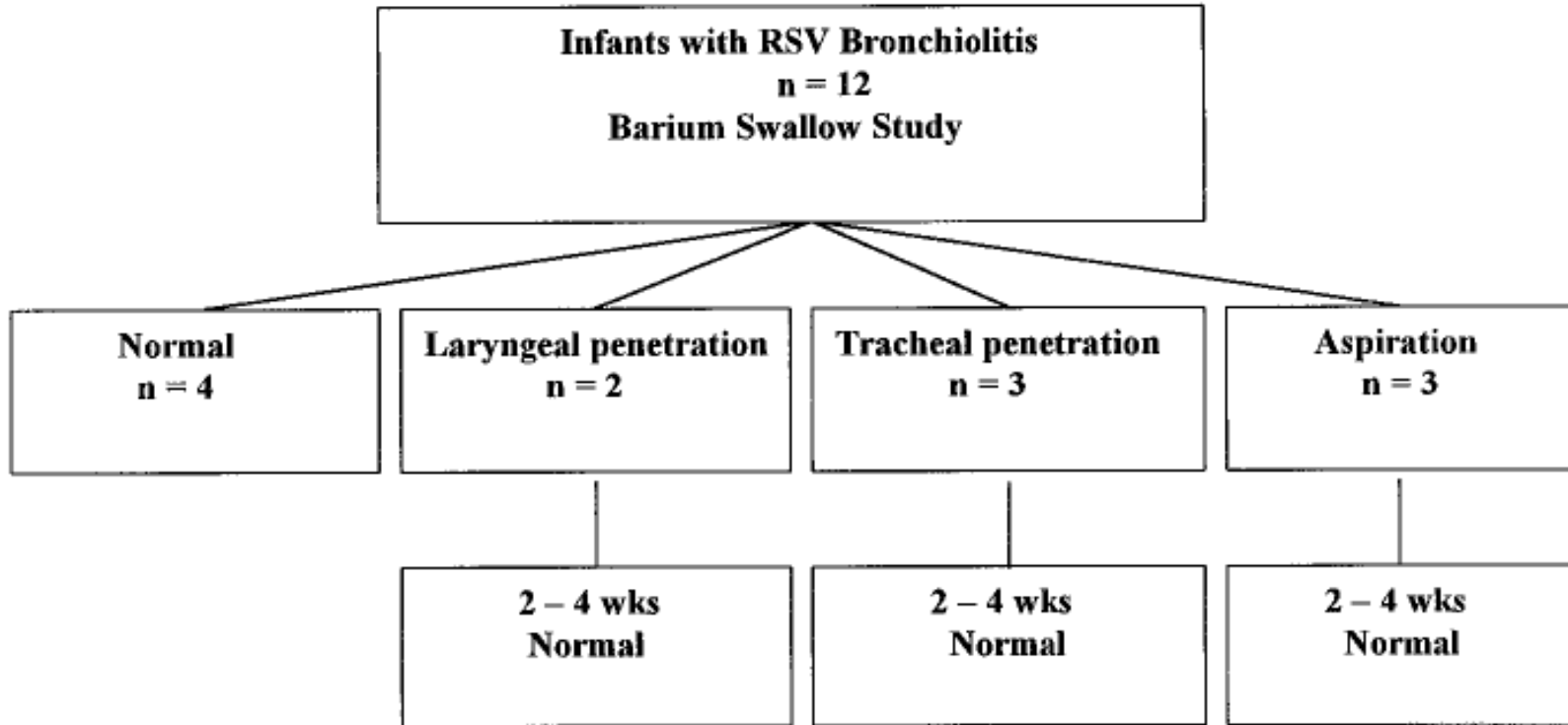
– Severity of lung disease

- Some infants/children become symptomatic with minimal aspiration while others can tolerate aspiration without significant lung injury

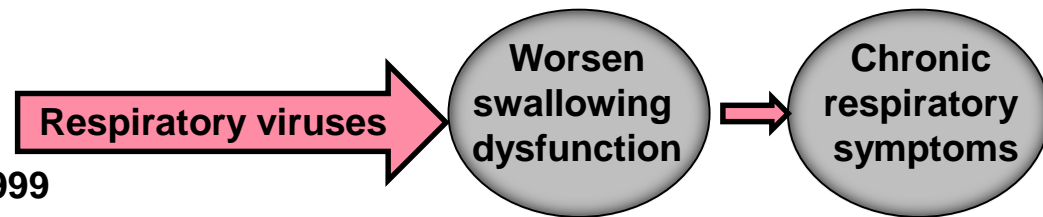
Treatment options for infants and children with swallowing dysfunction and respiratory symptoms

- **Avoidance of textures that cause aspiration** based on videofluoroscopic swallow study
- **Supplement with enteral gastric tube feedings**
 - Many children with aspiration and swallowing dysfunction can take a limited oral diet until swallow improves
- **Gastroesophageal reflux with swallowing dysfunction and aspiration**
 - Can try antireflux medications- H₂ blockers or proton blockers
 - Severe GER with aspiration often requires surgical intervention
 - Nissen fundoplication and gastric tube
 - Gastro-jejunal tube placement

Also remember -respiratory viruses may temporarily worsen swallowing and aspiration in infants



In vulnerable children



Lung function and mucociliary clearance

Bulbar dysfunction, neuromuscular weakness recurrent aspiration can impair lung function

- **Peak cough flows may be decreased** in individuals with bulbar dysfunction
- **Reduced airway flows and decreased expiratory muscle strength may impair the ability to clear airway secretions**
 - **Increasing risk and severity of lower airway illnesses with respiratory viruses**
 - **Need for annual influenza vaccine and pneumovax**

Therapies to improve mucociliary clearance

- Chest physiotherapy
 - Not expensive but requires a person trained in doing it
- Therapy vest
 - Tolerated well, simple to use but expensive
- Exercise
- Acapella
 - Required good respiratory strength and a good mouth seal
- Cough-assist device
 - Requires training and some coordination

Therapy vest



Cough assist device



Spirometry to measure lung function



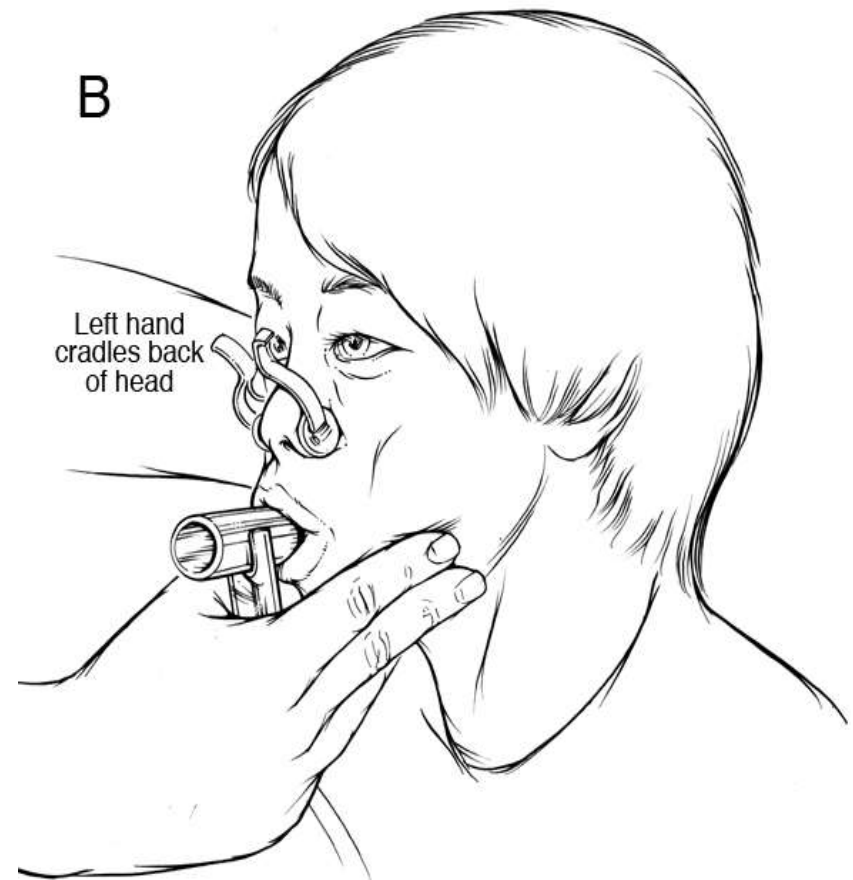
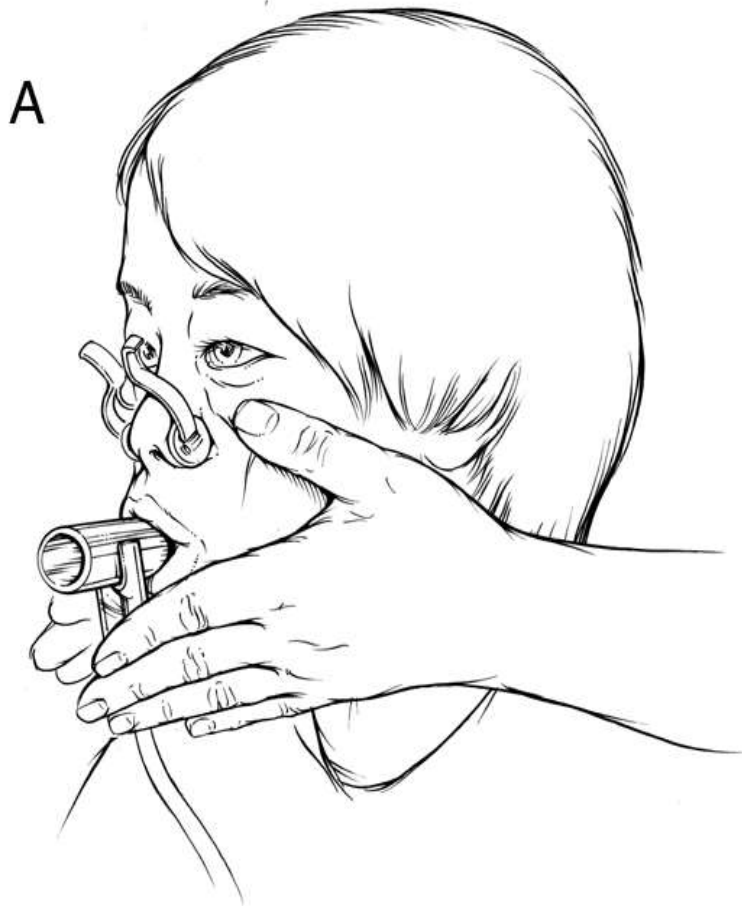
- Assess stability of lung function with age
- Assess response to therapy
- Children as young as 6yrs can perform spirometry

Force vital capacity (FVC) and FEV1

- FVC can drop with declining respiratory muscle strength
 - Decreased FVC associated with restrictive lung disease and worsening respiratory muscle strength
- Low FEV1% predicted can be associated with obstructive lung disease
 - Decreased **FEV1/FVC ratio**- seen in children with asthma and other airway diseases (recurrent aspiration)

Adjustments may be necessary to monitor lung function by spirometry

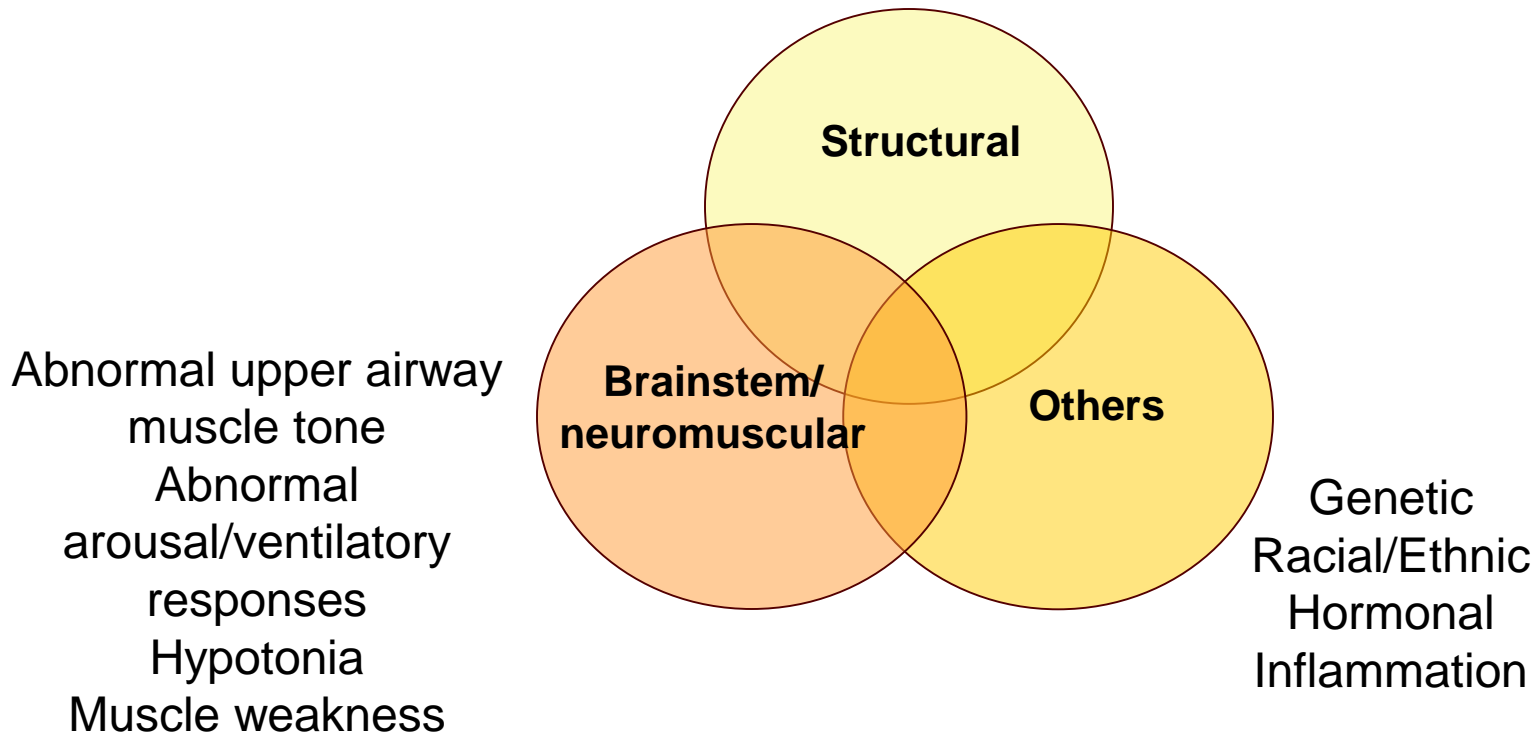
Optimizing head position and mouth-seal



Sleep disordered breathing and abnormalities in ventilation

Causes of sleep disordered breathing

Adenotonsillar hypertrophy, narrow pharyngeal structure,
craniofacial dysmorphology, obesity



Abnormalities in gas exchange

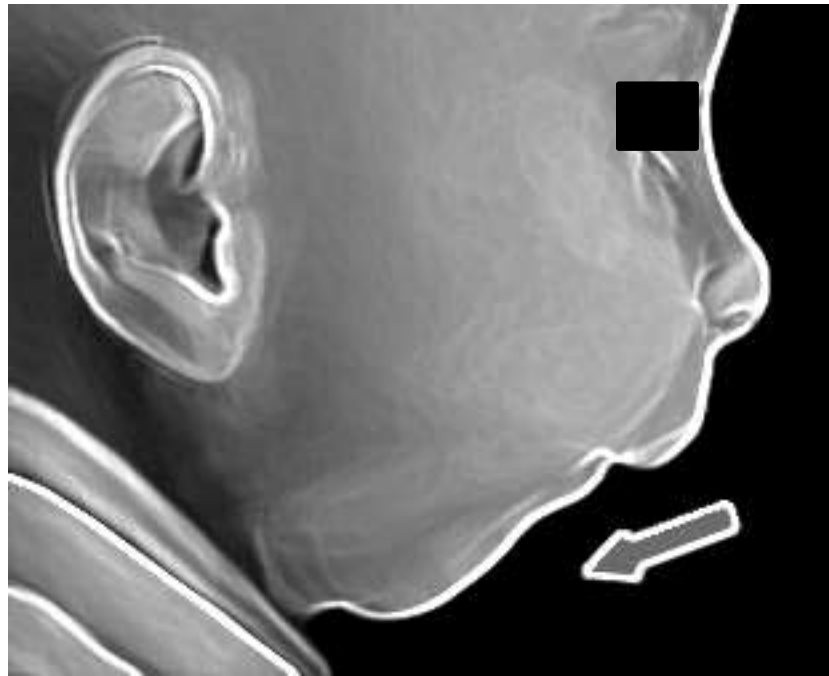
- Craniofacial, neuromuscular and brainstem abnormalities-
 - Can lead to upper airway obstruction, hypoxia and hypercarbia with sleep and during illnesses and stress
 - Central apneas and oxygen desaturations with brainstem abnormalities and decreased pulmonary reserve
 - Gas exchange abnormalities can be present at birth or develop with increasing age

Abnormalities in control of breathing

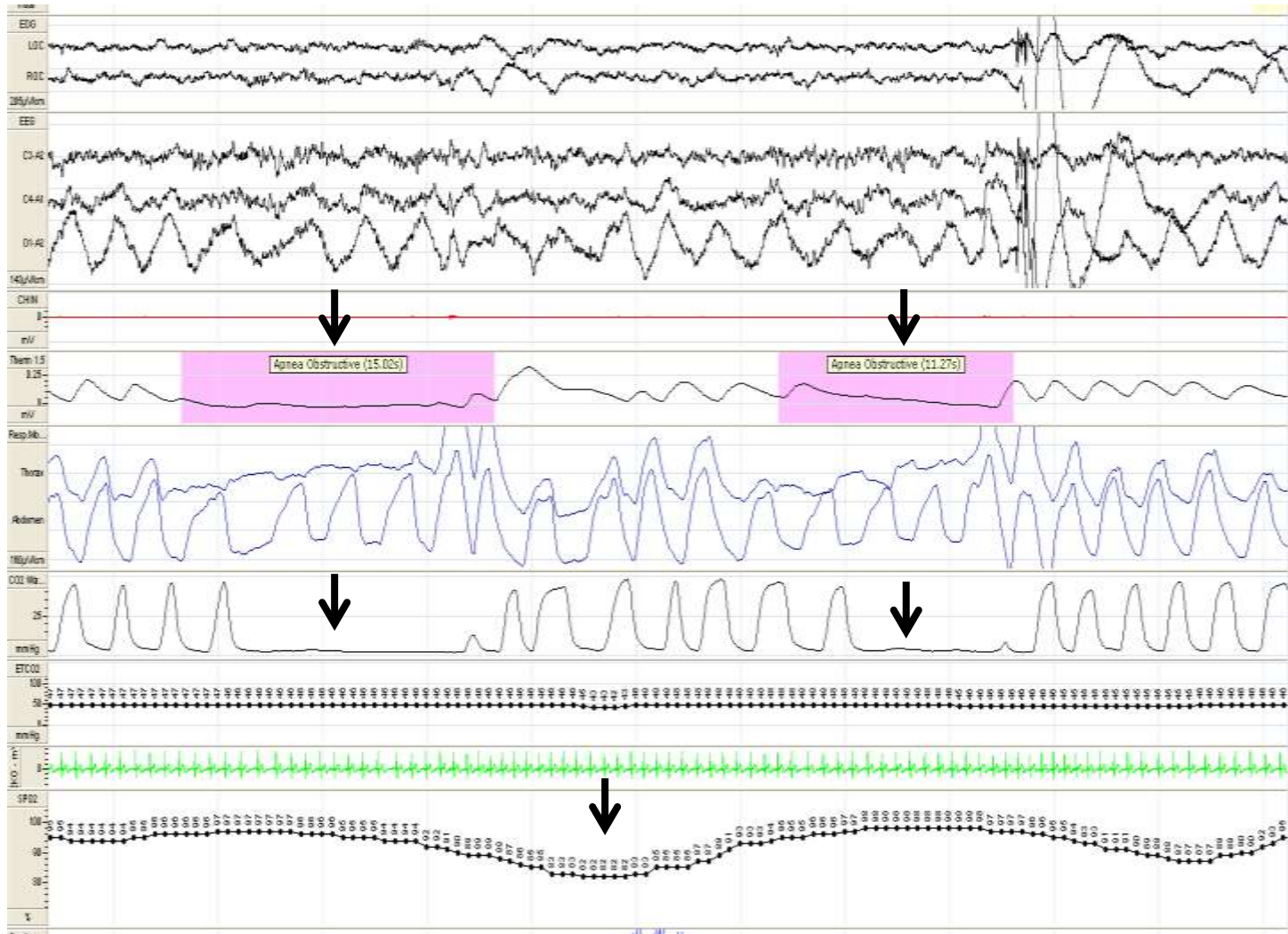
- Micrognathia may increase risk of upper airway obstruction
- Obesity and hypotonia may increase risk of obstructive sleep apnea
- Neuromuscular weakness may increase risk of partial obstructions
- Decreased pulmonary reserve from aspiration or recurrent lower airway illnesses may cause oxygen desaturations
- Brainstem abnormalities may increase risk of central apneas
- Combinations of the above

Structural airway abnormalities can cause obstructive apnea awake and asleep

Micrognathia



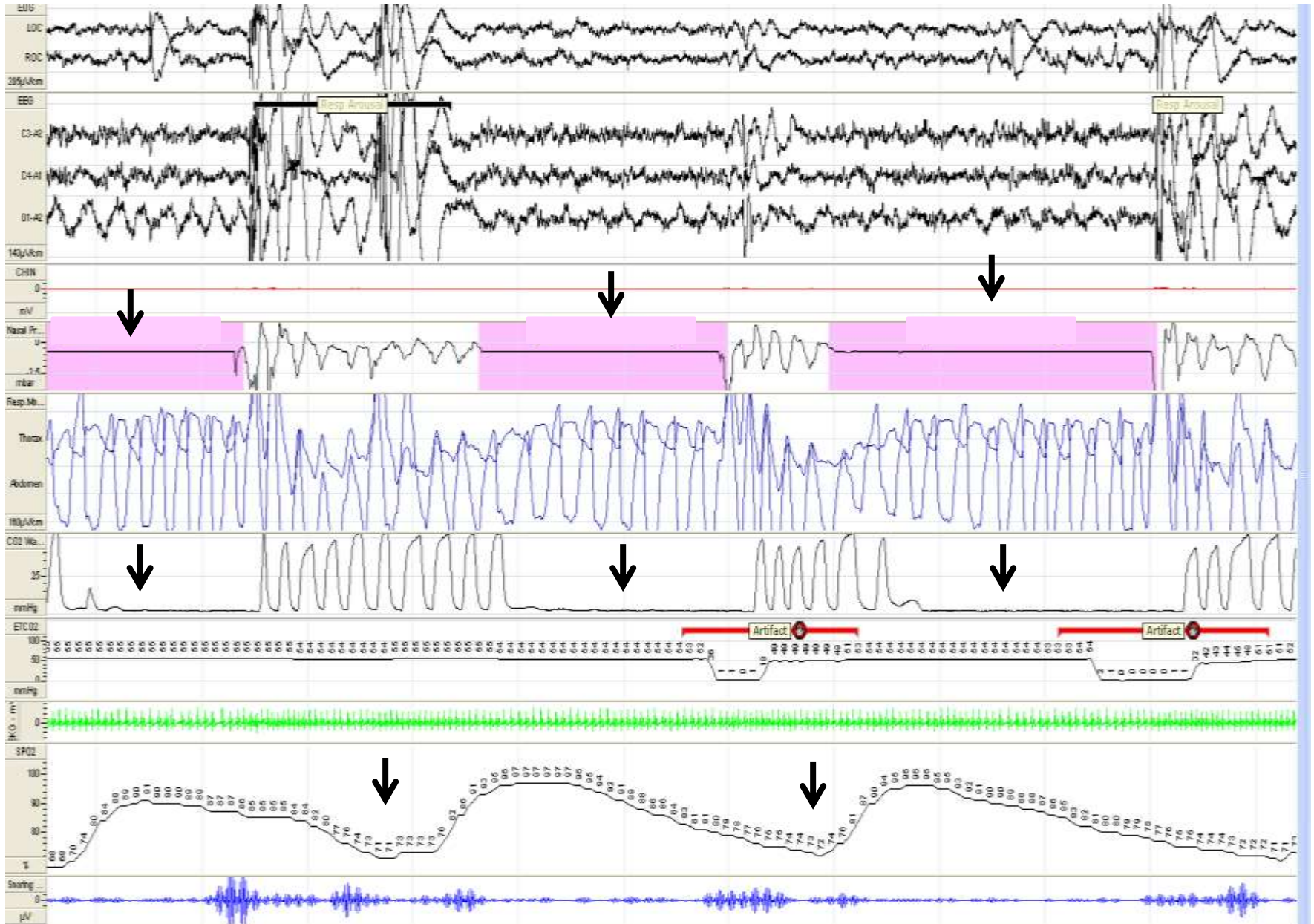
Obstructive apnea on sleep study



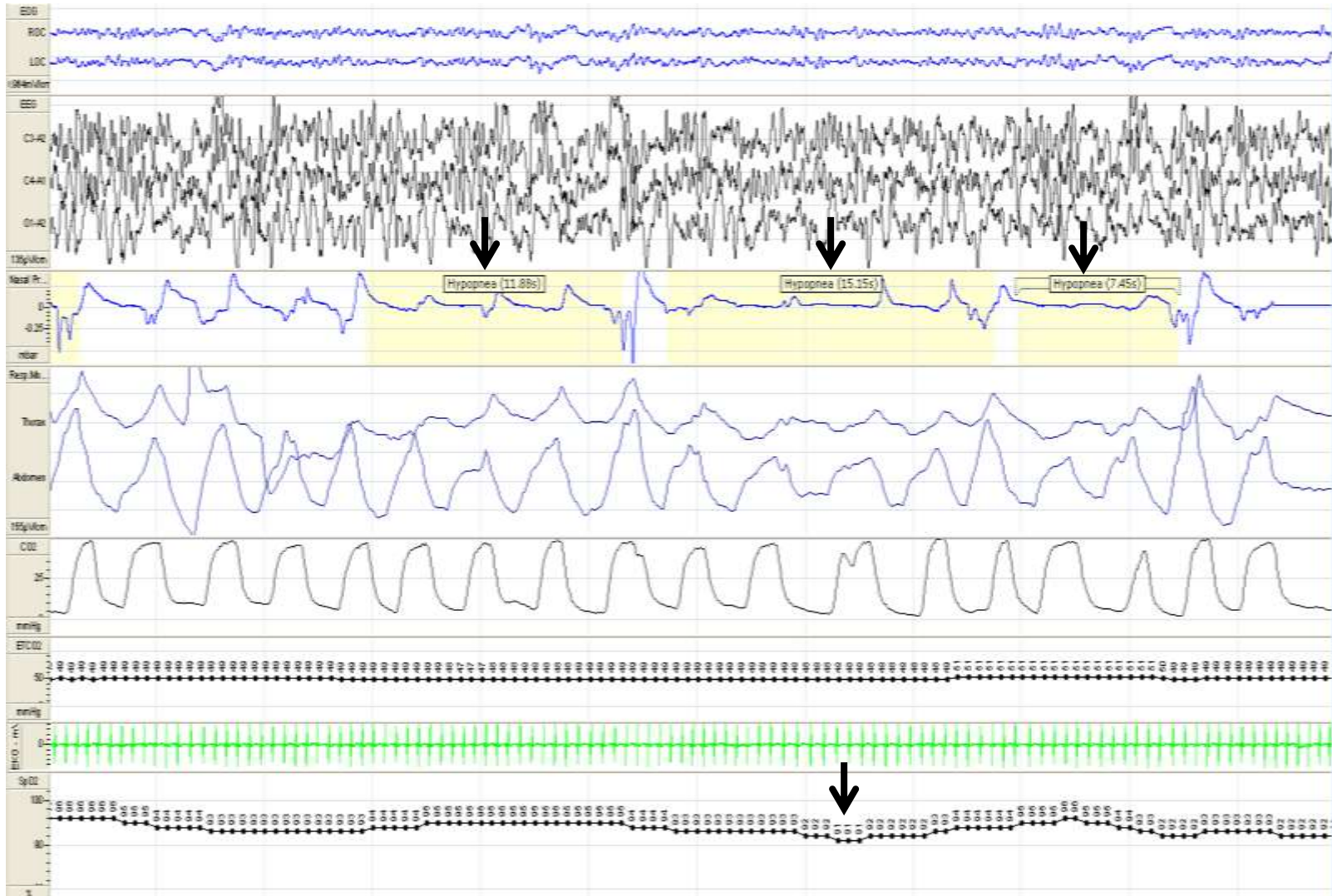
Obesity can cause obstructive apnea



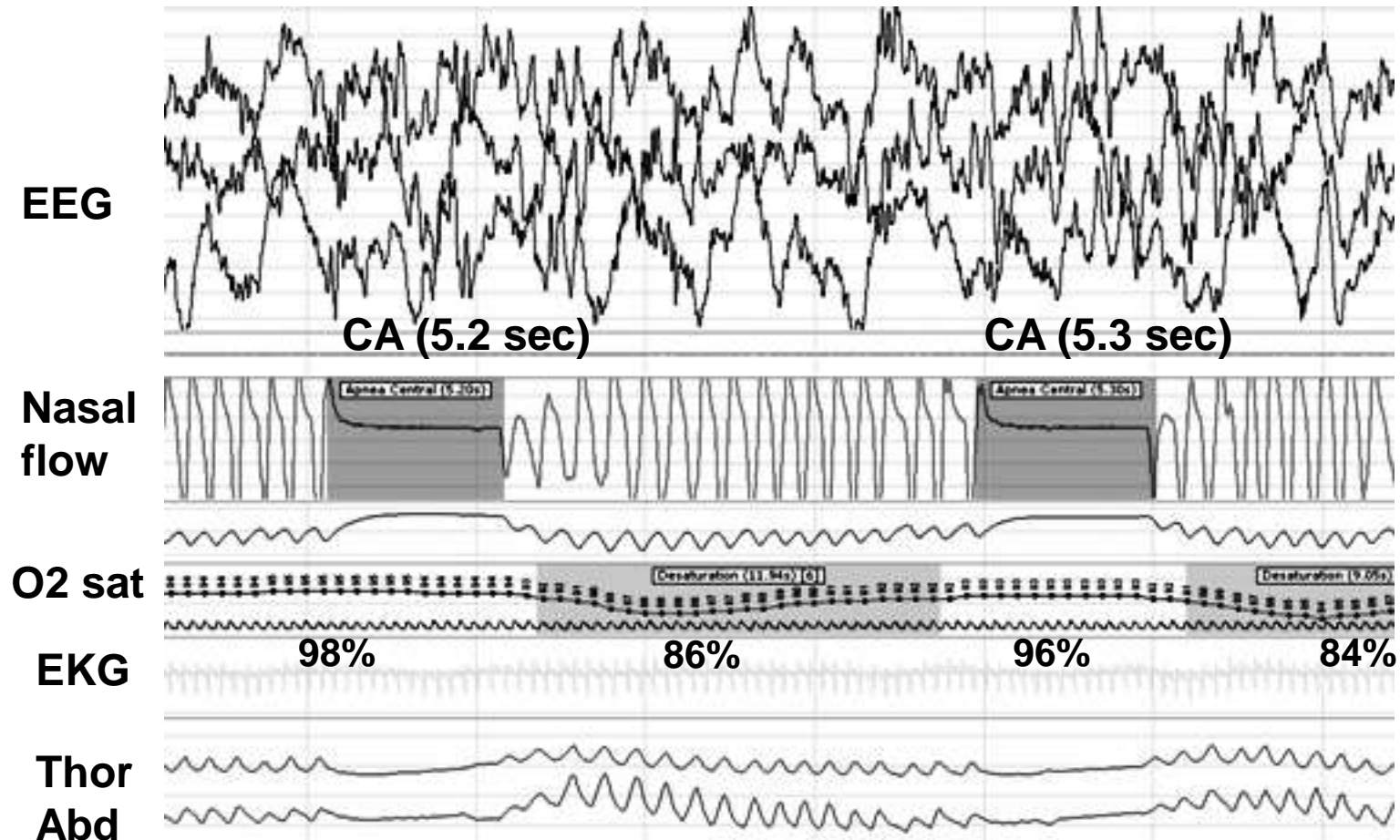
Obstructive apnea on sleep study from obesity



Neuromuscular weakness and hypotonia can cause partial upper airway obstruction and oxygen desaturations



Decreased pulmonary reserve from lower respiratory tract disease



Interventions/treatment used for control of breathing problems

- Micrognathia
 - ENT evaluation
 - Jaw distraction, tracheostomy, positioning, non-invasive ventilation (cpap/bipap)
- Obesity
 - ENT evaluation of tonsils, adenoid or other upper airway abnormality
 - CPAP
- Neuromuscular weakness and hypotonia
 - Non-invasive ventilation
- Central apneas due to brain stem abnormalities
 - Positive pressure ventilation and tracheostomy
 - Non-invasive ventilation

Nasal mask and CPAP device



Nasal pillows for non-invasive ventilation



Respiratory management of children with Mobius syndrome

- Monitor respiratory symptoms closely
- Respiratory symptoms can be worse in the very young, with illnesses and in children with co-morbidities
- Annual flu shots and consider pneumovax every 5-7 years in children at risk for more severe respiratory compromise
- Sleep studies to rule out obstructive or central apneas as indicated
- Swallow studies in child with symptoms of swallowing disorder or chronic respiratory symptoms
- Monitor lung function with age
- Aggressive chest physiotherapy and mucociliary clearance techniques as indicated