Respiratory management of neuromuscular disease in children

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Aetiology

- Central
  - Brainstem
  - Cerebral palsy
- Spinal
  - Trauma / tumour/ infection
- Nerve
  - Poliomysositis
  - myasthenia gravis
  - Spinal muscular atrophy
  - Neuropathy – hereditary and acquired (Guillain Barre)
  - Phrenic nerve injury

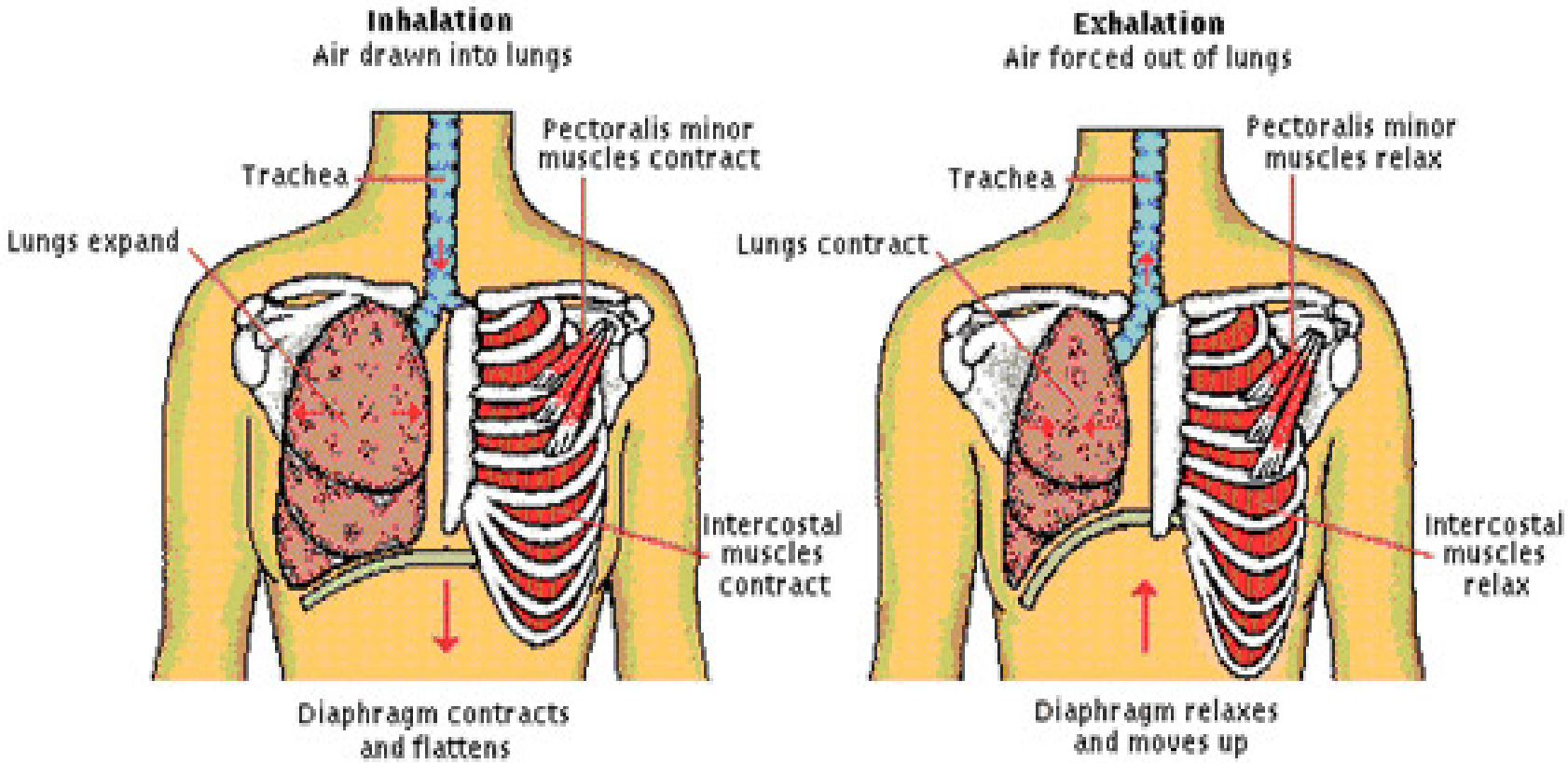
Aetiology

- Muscle
  - Muscular dystrophies
    - Duchenne/Becker
    - Limb-girdle
    - Emery-Dreifuss
    - Fascioscpulohumeral
    - Congential
    - Myotonic dystrophy
  - Myopathy – Nemaline
- Chest wall
  - Jeune syndrome (asphyxiating thoracic dystrophy

Duchenne muscular dystrophy

- X-linked recessive
- 1:3000 births
- Mutation of dystrophin gene
- Progressive loss of muscle strength
  - Loss of ambulation
  - Respiratory insufficiency

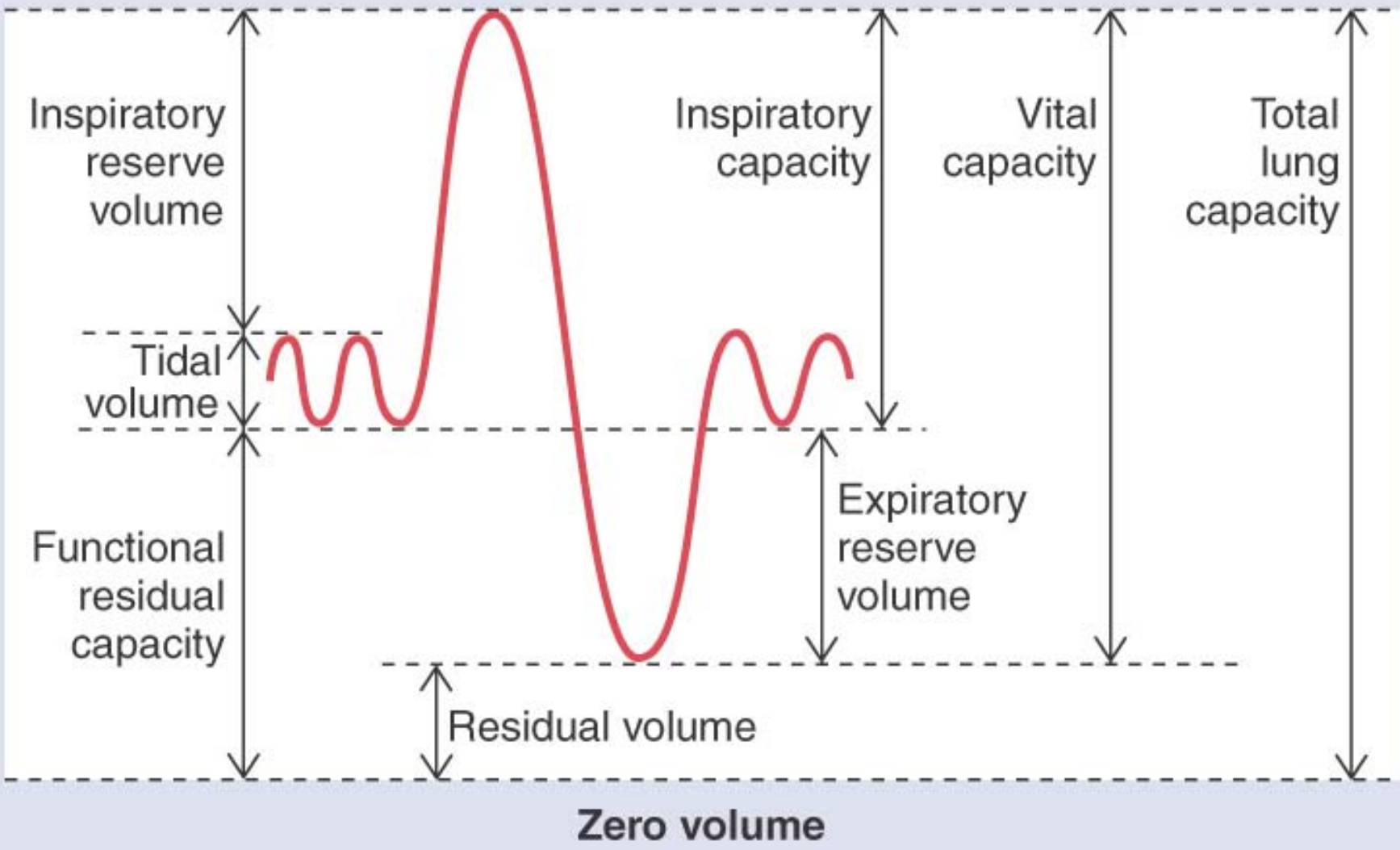
Respiratory muscle function



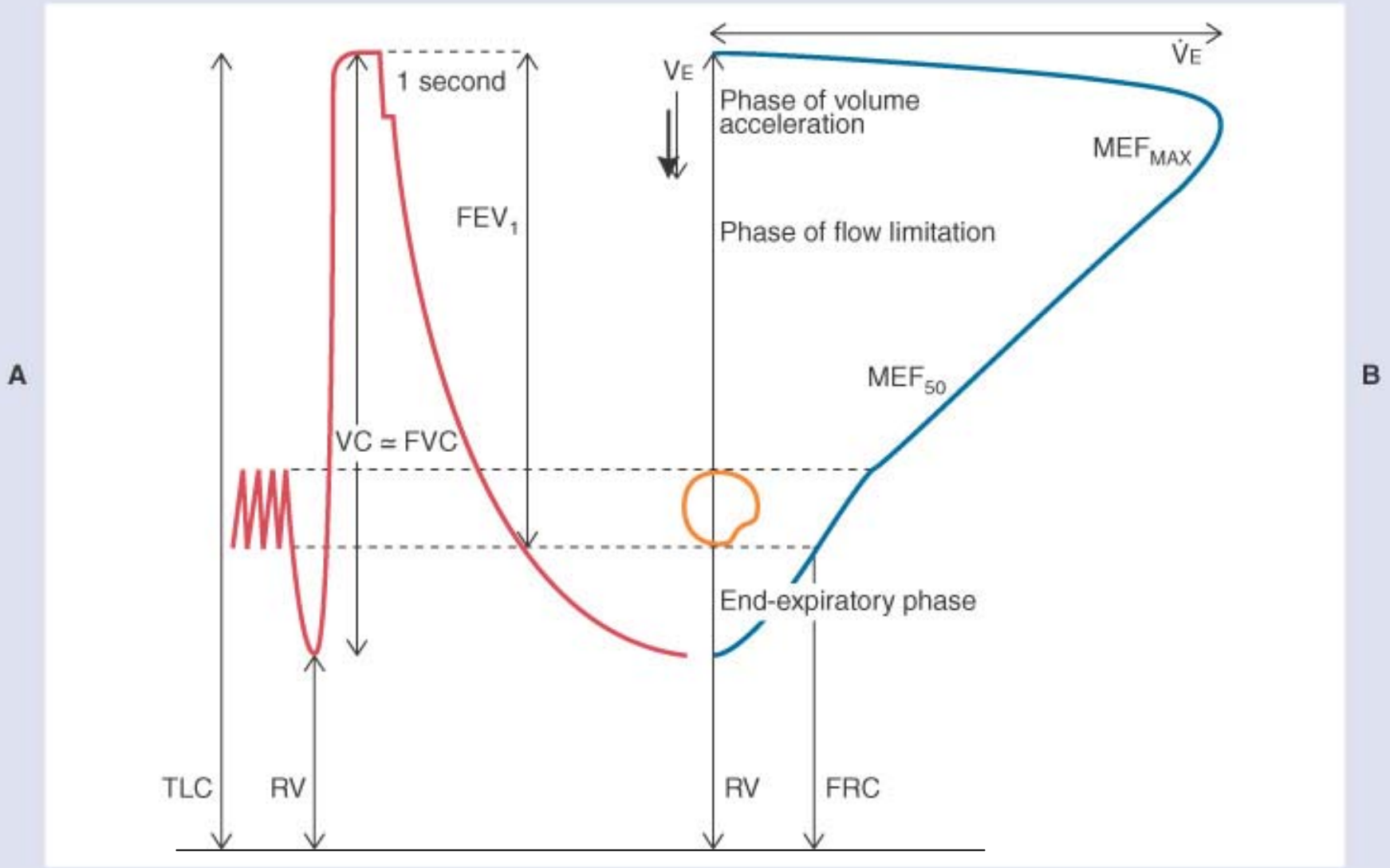
Lung function in neuromuscular weakness

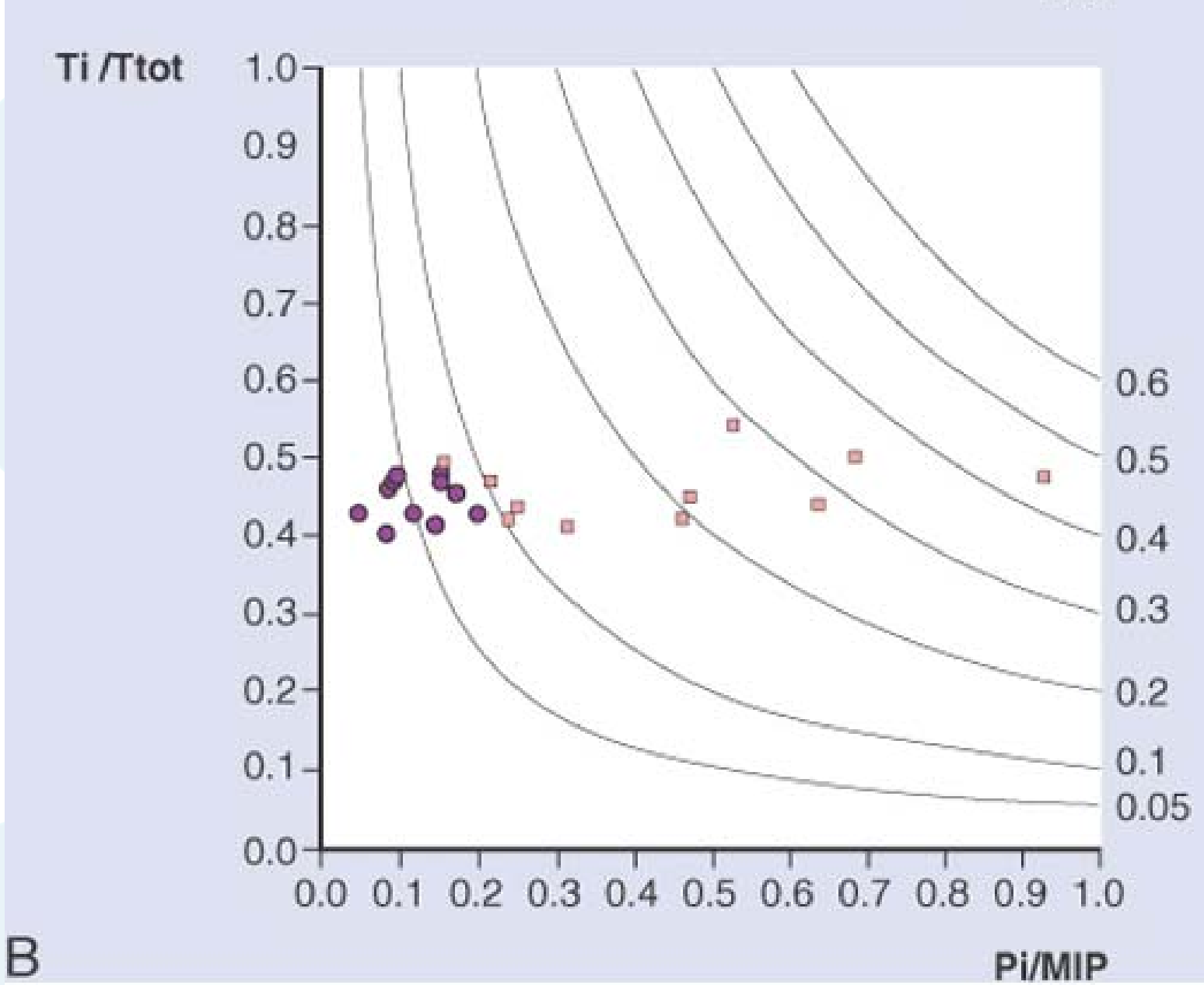
- Muscle
  - ⇓ inspiratory flow
  - ⇓ inspiratory capacity
  - ⇓ expiratory flow
  - Muscle fatigue
- Lung
  - ⇓ compliance
- Chest wall
  - ⇓ compliance
- Result
  - Restrictive defect (reduced breath volume)

Lung function testing



Lung function testing





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Tension Time index

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Lung function in neuromuscular weakness

- reduced total lung capacity
- reduced vital capacity
- reduced maximal flows

Chest wall alterations

- Compliance
  - Initially ⇑ compliance
  - Progress to ⇓ compliance
- Chest wall distortion - scoliosis
  - Reduced muscle efficiency

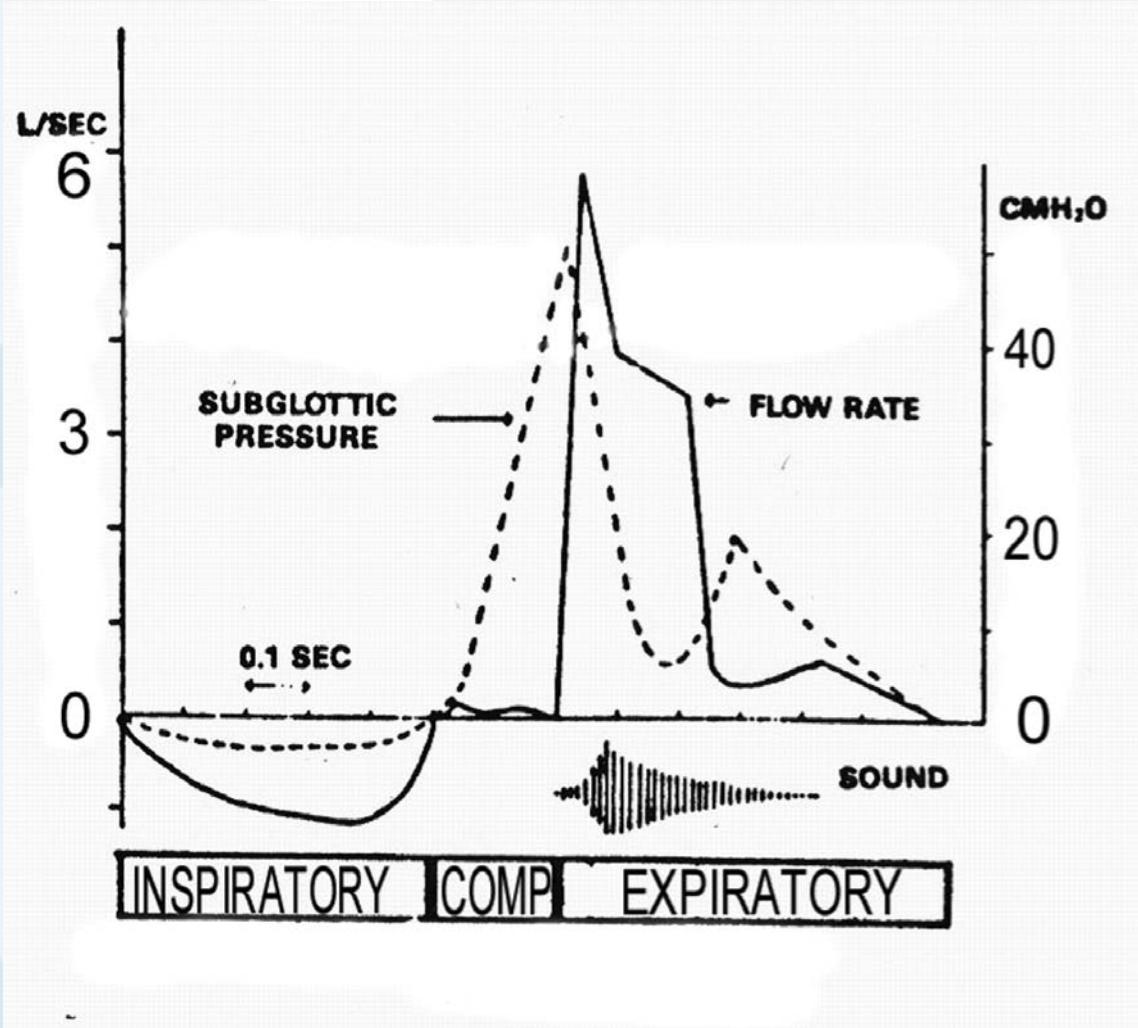
Control of breathing

- Adaptive impairment of control of breathing
  - CO_2 retention

Cough

- important for airway clearance
- dependent on good respiratory muscle strength
  - inspiratory and expiratory muscles

Schematic diagram depicting changes in flow and subglottic pressure during the inspiratory, compressive (COMP), and expiratory phases of cough



McCool, F. D. Chest 2006;129:48S-53S

Cough - impaired

- ⇓ inspiratory capacity
- ⇓ glottic closure
- ⇓ expiratory flow

- Result:
  - impaired airway clearance

Consequences

poor airway clearance
recurrent pneumonia
lobar collapse
dysphagia
aspiration
Malnutrition
scoliosis
nocturnal hypoventilation
respiratory failure
cor pulmonale

Impaired airway clearance

- recurrent pneumonia
- persistent lobar collapse
- bronchiectasis

Swallowing dysfunction

- dysphagia
- impaired glottic function
- gastro-oesophageal reflux
- aspiration
- malnutrtion

Obstructive sleep apnoea

- Aetiology:
  - Reduced upper airway tone
  - Reduced inspiratory muscle strength
  - Obesity
- Symptoms
  - snoring
  - early morning headaches/ lethargy
  - daytime somnolence
- Diagnosis
  - sleep study

Nocturnal hypoventilation

- REM related hypoventilation
  - reduced intercostal muscle function
  - Sleep fragmentation
- Symptoms
  - early morning headaches / lethargy
  - daytime somnolence
  - learning impairment
- Diagnosis
  - sleep study
  - early morning CO2

Respiratory failure

- Progression from nocturnal hypoventilation
- Progress to cor pulmonale

- Cardiomyopathy

Management

- recognition of symptoms
- regular review
- anticipation of complications
- early intervention

Airway clearance

- maintain mobility
- encourage deep breathing
- bubble PEP
- assisted coughing
- Cough Assist

Cough Assist

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Cough Assist

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Nutrition

- excess nutrition
  - reduced mobility
  - impaired lung function
- inadequate nutrition
  - poor swallow
  - reduced muscle strength
  - consider supplemental nutrition / gastrostomy

Nocturnal hypoventilation

- recognition
- non-invasive positive pressure ventilation (NIPPV)
- improves ventilation during sleep
- improves quality of sleep
- improves daytime functioning
- prolongs survival

Non invasive positive pressure ventilation

Scoliosis

- results from muscle weakness
- distorts the chest cage
  - impairs muscle efficiency
  - reduced lung volume
- stabilised with surgery
  - pre operative evaluation
  - post operative physiotherapy / ventilatory support
  - does not reduce rate of decline in lung function

Regular reviews

- lung function
- swallowing
- sleep study
- cardiac assessment
- airway clearance techniques
- orthopaedic assessment
  - scoliosis

Consequences of prolonged survival

- Inevitable:
  - Progressive respiratory failure
  - cardiomyopathy
- Expected
  - Progressive dysphagia
  - Malnutrition
  - Osteopenia / fractures
  - Social / burden of care
- Unexpected
  - Nephrolithiasis
  - Diabetes mellitus
  - Deep vein thrombosis
  - Gall stones

End of life decisions

- quality of life
  - informed participation of patient and family
- clearly documented
- continuous invasive ventilation

Multidisciplinary Clinic

- Neurologist
- Respiratory Physician
- Orthopaedic Surgeon
- Physiotherapist
- Dietician
- Social Worker
- Occupational Therapist
- Clinic Coordinator

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Thank you

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