Neuromuscular Conditions

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http://rc-edconsultant.com/

Learning Objectives:
- Describe the pathophysiology, manifestations, diagnosis and general management for the following conditions:
  - myasthenia gravis
  - Guillain Barre syndrome
  - amyotrophic lateral sclerosis
  - muscular dystrophy
  - spinal muscle atrophy
  - critical illness polyneuropathy and myopathy
- Describe techniques for assessment of weakness of ventilatory apparatus including physical assessment, lung volume measurement, blood gases, inspiratory and expiratory pressures and sleep studies.

Learning Objectives:
- Describe techniques applied to non-ventilatory management of neuromuscular conditions.
- Identify the goals for mechanical ventilation for patients with neuromuscular disease.
- Describe the selection and application of mechanical ventilation methods to neuromuscular conditions.

Neuromuscular Pathophysiology

Requirements for ventilation
- neural ventilatory drive - stimulus to breathe
- neural transmission to muscles of ventilation
- contractility of ventilatory muscles
- feedback to CNS; e.g., from chemoreceptors

Requirements for ventilation
- receptors- input to ventilation center
  - brainstem- pH sensor
  - carotid bodies- oxygen
  - lung stretch receptors - mechanics
Requirements for ventilation
- Ventilatory drive - centers in medulla and pons (brainstem)
  - Medullary ventilatory center - regularity of pattern
  - Pneumotaxic & apneustic centers - frequency and tidal volume

Requirements for ventilation
- Nerve transmission from centers to ventilation muscles
  - Phrenic nerve - from C3-C5
    - Innervates diaphragm
  - Intercostal nerves - from T1-T12
    - Innervate intercostal muscles

Requirements for ventilation
- Nerve transmission from centers to ventilation muscles
  - Abdominal nerves
    - Thoracic and lumbar spine
    - Innervate abdominal muscles

Requirements for ventilation
- Muscular contraction
  - Transmission across myoneural synapse
  - Muscular function

Neuromuscular impairment of ventilation
- CNS (brain, spinal cord) failure
  - Trauma
  - Infection
  - Inflammation
  - Toxins
  - Drugs
  - Ischemia/hypoxia
  - Congenital dysautonomia - inborn failure of breathing automaticity

Neuromuscular impairment of ventilation
- Nerve transmission failure
  - Trauma - transection of spinal cord
    - C1-C3 - absence of nerve transmission to all ventilatory muscles
    - C6-C7 - absence of transmission to intercostal & abdominal muscles
**Neuromuscular impairment of ventilation**
- nerve transmission failure
  - iatrogenic - laceration of phrenic nerve
    - occurs during heart surgery
    - may self-reverse (months)
  - infection
    - poliomyelitis
    - West Nile virus
- degenerative disease
  - amyotrophic lateral sclerosis
  - multiple sclerosis
- autoimmune disease
  - Guillain-Barre syndrome
  - myasthenia gravis
  - Lambert-Eaton syndrome

**Neuromuscular impairment of ventilation**
- nerve transmission failure
  - toxins - botulism (neuromuscular junction)
  - drugs
    - neuromuscular blockers
    - anticholinesterase agents

**Neuromuscular impairment of ventilation**
- muscular failure (congenital)
  - spinal muscle atrophy
  - muscular dystrophy

**Neuromuscular impairment of ventilation**
- muscular failure (acquired)
  - polymyositis (inflammation)
  - atrophy; e.g., ventilator-induced diaphragmatic dysfunction (VIDD)
  - fatigue - excessive WOB
  - rhabdomyolysis - breakdown of muscle
  - critical illness polyneuropathy

**Components of ventilatory impairment**
- Inspiratory muscle weakness - decreases lung volumes
- Expiratory muscle weakness - decreases expiratory flow (cough)
- Bulbar weakness - muscles of:
  - throat
  - jaw
  - tongue
  - face
  - predisposes to aspiration
Electrophysiology Tests

Purposes

- Confirm presence of neuromuscular disorder
- Distinguish between nerve, muscle and neuromuscular junction disorders
- Identify specific diagnosis

FYI - Link to RC article that explains electrophysiology testing
http://www.rcjournal.com/contents/09.06/09.06.1024.pdf

Types

- nerve conduction studies
- needle electromyography
- neuromuscular junction testing
- repetitive nerve stimulation
- single-fiber EMG
- train-of-four stimulation
- phrenic nerve conduction
- needle EMG of diaphragm

Nerve conduction studies

- Defined - percutaneous nerve stimulation with surface recording of conduction

FYI - Click for article on nerve conduction studies

Nerve conduction studies Parameters

- compound muscle action potential (CMAP) - sum of the response of all stimulated muscle fibers
- conduction velocity - if decreased, then neuropathy is present
- F wave - absence indicates demyelination, impairs conduction

Needle electromyography

- Needle inserted in muscle and measures motor unit potentials (MUP) with voluntary contractions
- Decreased duration/amplitude of motor unit potential suggests a myopathy

Click to see images of needle EMG
http://www.teleemg.com/new/atlas.htm
<table>
<thead>
<tr>
<th>Repetitive nerve stimulation</th>
<th>Train-of-four stimulation</th>
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<tbody>
<tr>
<td>Surface electrodes produce repetitive, strong stimuli</td>
<td>Used to titrate surgical neuromuscular blockers</td>
</tr>
<tr>
<td>Early decrement in compound muscle action potential (CMAP) indicates neuromuscular junction disorder; e.g., myasthenia gravis</td>
<td>Four stimuli of ulnar nerve, with observation of thumb twitches - desired is 1 or 2 twitches per 4 stimuli</td>
</tr>
</tbody>
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Click for more information on train-of-four testing [http://www.globalrph.com/neuromuscular.htm#Train_of_four](http://www.globalrph.com/neuromuscular.htm#Train_of_four)

<table>
<thead>
<tr>
<th>Phrenic nerve conduction</th>
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<tbody>
<tr>
<td>Technique</td>
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<tr>
<td>- Surface electrodes, bilaterally on neck stimulate phrenic nerve</td>
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<tr>
<td>- Diaphragm CMAP measured with electrodes placed at the xiphoid process and costal margin</td>
<td>- Diaphragm CMAP measured by electrodes at xiphoid process and costal margin</td>
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<tr>
<td>Significance</td>
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<tr>
<td>- bilateral decreased amplitude - neuropathy</td>
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<tr>
<td>- unilateral decreased amplitude - traumatic or surgical lesion</td>
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<tr>
<th>Guillain-Barre Syndrome</th>
<th>Description &amp; demographics</th>
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<td>Group of five autoimmune peripheral neuropathies</td>
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<tr>
<td>- acute inflammatory demyelinating polyneuropathy (90% of cases)</td>
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<td>- acute motor axonal neuropathy (young people)</td>
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<td>- acute motor sensory axonal neuropathy (uncommon)</td>
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<tr>
<td>- Miller-Fisher syndrome (rare)</td>
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<tr>
<td>- acute pandysautonomia (rarest)</td>
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Epidemiology
- Most common neuromuscular condition for ICU admission
- 1-3 cases/100,000/year
- All age groups
- No gender preference
- Incidence unrelated to current influenza vaccines

Pathophysiology
- Antecedent infection - one to three weeks before GBS onset
- Interaction of pathogen and nerve tissue activates autoimmune response - immunity to self

Pathophysiology
- Antecedent infection - one to three weeks before GBS onset
- Interaction of pathogen and nerve tissue activates autoimmune response - immunity to self
- Autoimmune response produces antibodies that cause demyelination of nerve cells
- Demyelination impairs neural conduction

Common pathogens
- campylobacteriosus - most common
- cytomegalovirus
- Epstein-Barr virus
- varicella zoster virus
- mycoplasma pneumoniae

Manifestations
- Severity is variable - weakness to total paralysis with autonomic dysfunction
- Evolves over hours to days
- Ascending paralysis - starts as 'rubbery legs'
- Tingling in extremities

Manifestations
- Does not affect consciousness - patients are alert and frightened
- May involve cranial nerves - bulbar weakness
- Loss of deep tendon reflexes
- Pain in affected muscles; sometimes back pain
- Ventilatory failure in 30% of patients
Differential diagnosis
- Myasthenia gravis
- Botulism
- Polymyositis
- Poliomyelitis
- West Nile virus
- Heavy metal intoxication; e.g., arsenic
- Tick paralysis (Lyme disease)
- Porphyria - a hemoglobin disorder

Diagnosis
- Clinical manifestations; e.g.:
  - ascending paralysis
  - areflexia
  - pain
- Laboratory - elevated CSF proteins; 48H after onset

Diagnosis
- Electrodiagnostic features
  - may be absent in early stages
  - decreased nerve conduction velocity
  - needle EMG - abnormal spontaneous activity
  - normal muscle response to direct stimulation

Management
- Supportive
  - respiratory care
  - nursing care
  - psychological, emotional support

Management
- Intravenous immunoglobulin (IVIg)
  - neutralizes antibodies
  - five daily infusions
- Plasmapheresis
  - blood removed from patient, plasma separated, then treated to remove antibodies, then returned or replaced with substitute fluid
  - complications (uncommon)
    - hypotension
    - hemorrhage
    - sepsis - immunosuppression

Click to see picture of patient on plasmapheresis
http://www.flickr.com/photos/dunegod00/2526944266/in/photostream/
### Management
- Plasmapheresis vs. IVlg
  - They are equally effective for GBS
  - IVlg easier to administer
  - IVlg has fewer complications

### Course, prognosis
- Worst clinical function usually in first week
- Nearly full function at four weeks
- Full recovery within months for 85% of patients
- Relapse occurs in 5-10%
- Mortality <5%

## Myasthenia Gravis

### Description
- Autoimmune disorder that compromises transmission across the neuromuscular junction.
- Characterized by weakness and fatigability of striated muscles.
- Broad range of severity - from drooping eyelids (ptosis) to ventilatory failure.

### Epidemiology
- 14 cases per million in US (not rare)
- Occurs in all age groups
- Females, peak occurrence - 20-40 YO
- Males, peak occurrence - 40-60 YO
- Congenital form - onset in utero, with decreased fetal movement
- Juvenile form - onset in 20s

### Pathophysiology
- Autoimmune activity against acetylcholine receptors by anti-AchR antibodies
Nerve impulse $\implies$ release of ACh $\implies$ AChR $\implies$ depolarization of muscle

Release of ACh-ase $\implies$ hydrolyzes ACh $\implies$ repolarization of muscle for next contraction

Myasthenia gravis - AChR antibodies decrease available receptors, preventing depolarization

Manifestations
- Does not affect consciousness
- Weakness, initially in ocular muscles
  - ptosis (drooping eyelids)
  - diplopia (double vision)
- Difficulty chewing, speaking, singing, swallowing
- Fatigability - improves with rest
- Descending paralysis

Differential diagnosis
- Hypothyroidism
- Medications may induce or exacerbate MG:
  - antibiotics; e.g., aminoglycosides
  - cardiovascular agents; e.g., propanolol
  - anti-malarial agents; e.g., chloroquine
  - miscellaneous agents
- Hypothyroidism
- Adverse effects of medications
- Amyotrophic lateral sclerosis
- Botulism - strong resemblance
- Guillain-Barre syndrome
- Polymyositis
- Lambert-Eaton syndrome - complication of carcinoma
- Multiple sclerosis

Click to see video of MG patient detained by police (2 min)
http://www.youtube.com/watch?v=ypwjG4J7JKM
### Diagnosis

- **Clinical presentation**
  - Ice pack test - ice improves eyelid fatigue
  - Tensilon (edrophonium HCl) test - anticholinesterase agent increases ACh, increasing ACh-receptor interactions
    - **onset** 30 sec.
    - **duration** 5 min.
  
  Click to see video of positive Tensilon test
  [http://www.youtube.com/watch?v=k7YX9kuWrxA](http://www.youtube.com/watch?v=k7YX9kuWrxA)

- **Electrophysiology - repetitive nerve stimulation**
  - rapid decline in amplitude of induced responses
  - specific for neuromuscular junction disorders

- **Laboratory - AChR-Ab**
  - specific for MG
  - false negative results in 50% of MG patients who have only ocular weakness

- **Medical imaging - chest CT with contrast** is indicated
  - 20% MG patients have thymoma
  - 70% MG patients have thymic hyperplasia

Click to see thymus enlargement with MG

### Management

- **Supportive - severe weakness** (myasthenic crisis) may require intubation, ventilation

- **Evaluate for thymectomy**
  - improvement (delayed) in 85%
  - remission in 35%
  - recommended for all MG patients between puberty and 55 YO
  - Thymectomy - should be done in center with experience
Management

- Anticholinesterase agents
  - oral pyridostigmine (Mestinon)
  - symptomatic improvement
  - bedside pulmonary function testing by RTs for dosage, duration of action

Management

- Cholinergic crisis - side effect, due to overdose with pyridostigmine
  - signs
    - bradycardia
    - bronchospasm
    - excessive secretions
  - management - atropine

Management

- Immunosuppression
  - severe weakness
    - plasmapheresis
    - IVIg
  - moderate weakness
    - glucocorticoids - improvement in most patients
    - azathioprine
    - mycophenolate mofetil
    - cyclosporine

Amyotrophic Lateral Sclerosis

Description

- AKA 'Lou Gehrig's disease'
- Devastating progressive neurodegenerative condition, causing:
  - painless weakness
  - muscular atrophy

FYI - Click to see Lou Gehrig video (.5 min)
http://www.youtube.com/watch?v=_LFprYdL2_6

Epidemiology

- Most common progressive neurodegenerative disease
- Incidence - 2 cases /100,000 people
- 5-10% of cases are inherited as autosomal dominant trait - familial ALS
- Male:female = 1.4:1
- 90% of ALS patients > 40 YO
- 5% of ALS patients < 30 YO
Epidemiology
- Possible risk factors:
  - smoking
  - oxidative stress
  - head injury - soccer players

Pathophysiology
- Death of motor neurons:
  - anterior horn cells
  - brainstem motor neurons
  - corticospinal motor neurons
- Denervation causes muscular atrophy
  (amyotrophy)

Click to see image of diseased spinal neuron
http://www.thehealthsuccesssite.com/images/muscle-als.jpg

Click to see image of ALS pathology
http://www.alsa.org/research/article.cfm?id=824

Manifestations
- Does not affect sensation or consciousness
- Manifestations vary, depending on which neurons are initially involved
  - lower neurons - limbs
  - bulbar neurons - face, tongue

Manifestations
- Does not affect sensation or consciousness
- Manifestations vary, depending on which neurons are initially involved
- Weakness
  - slow onset
  - asymmetric
  - starts distally in one limb
  - can start anywhere, progressing to everywhere

Manifestations
- Cramping
- Fasciculation (twitching)
- Exaggerated motor expressions of emotion (laughing, crying)
- Progressive paralysis
- Difficulty swallowing
- Ventilatory failure

Prognosis
- Common - death within 3 years of onset
- 20% survive 5 years
- 10% survive >10 years
Differential diagnosis

- Importance - ALS has no cure; so, alternatives that may be treatable must be ruled out before accepting ALS as diagnosis.

Differential diagnosis

- Other motor neuron diseases
- Structural disorders; e.g., CNS tumor, CNS radiation injury
- Toxic disorders; e.g., heavy metals
- Immune, inflammatory disorders; e.g,
  - multiple sclerosis
  - myasthenia gravis

Differential diagnosis

- Parkinson disease
- Thyrotoxicosis
- Infections; e.g.,
  - Lyme disease
  - syphilis
  - Creutzfeldt-Jakob disease

Diagnosis

- Clinical presentation
  - progressive course of weakness
  - hyperreflexia in weak, atrophied extremity
- Electrodiagnostics
  - denervation
  - fasciculation potentials

Management

- Supportive
  - respiratory care
  - nutrition
  - physical therapy
  - nursing
  - end-of-life care

Management

- Riluzole
  - glutamate antagonist
  - extends life by 2 months (average)
- Stem cell therapy - research in early stages
Muscular Dystrophy

Description
- Muscular dystrophy - a group of hereditary, progressive conditions that cause muscle fiber degeneration.

Description
- Duchenne muscular dystrophy (DMD)
  - most common
  - X-linked recessive trait
  - males only

Description
- Becker MD
  - second most common
  - milder than DMD
  - X-linked recessive - males only
  - Limb girdle MD - males & females
  - Emery-Dreifus MD - males & females

Epidemiology - DMD
- 1 case per 3500 live male births
- 1/3 of cases are spontaneous mutations ==> mom was not born with the defective gene

Pathophysiology
- Genetic defect for production of dystrophin
  - needed for muscle cell membrane integrity
  - deficiency permits leakage of muscle cell components ==> muscle cell death

Click to see video on histopathology of MD (3 min.)
http://www.youtube.com/watch?v=xwS3FtmvlRk
Manifestations

- Usual onset of frequent falling at 3-5 YO
- Gait abnormalities; e.g., toe walking
- Gower sign - arising from sitting position by going prone
- Inability to ambulate 7-13 YO
- Contractures

Progressive ventilatory failure

- Muscle weakness
- Kyphoscoliosis - restrictive lung dx
- Recurrent pulmonary infections

Other manifestations

- Progressive ventilatory failure
- Muscle weakness
- Kyphoscoliosis - restrictive lung dx
- Recurrent pulmonary infections

Cardiomyopathy - major cause of death

Cognitive deficit - IQ about 85

Death before 30 YO, due to cardio-pulmonary failure

Diagnosis

- Clinical presentation
  - Frequent falling
  - Gait abnormalities
  - Gower sign
- Electromyography - decreased action potential amplitude

Laboratory studies

- Elevated creatine phosphokinase (CPK) - DMD specific
- Muscle biopsy for histology
- Gene testing
  - Prenatal diagnosis
  - Carrier identification

Management

- Supportive
  - Respiratory care - lots of it
  - Nutrition
  - Physical therapy
  - Nursing
  - End-of-life care
Management

Special problem - kyphoscoliosis renders chest imaging difficult
- standard chest radiographs - difficult to position and interpret
- echocardiography - poor images

Click to see chest radiograph of severe kyphoscoliosis
http://www.scoliosisjournal.com/content/figures/1748-7161-2-15-5-i.jpg

Management

- Cardiac care
  - problems
    - cardiac dysfunction is masked by other limitations
    - dysrhythmias
    - ventricular failure
    - thromboembolic events

Management

- Cardiac care
  - early cardiac evaluation
  - diuretics, as indicated
  - angiotensin converting enzyme (ACE) inhibitors, as indicated
  - beta blockers, as indicated
  - anticoagulants, as indicated

Management

- Surgical procedures
  - surgical release of severe contractures
  - spinal fusion for kyphoscoliosis

Management

- Steroids - prednisone
  - prolongs walking
  - improves cardiac function
  - but -- side effects

Management

- Interventions under study
  - PTC 124 (Ataluren)
    - interferes with expression of defective genes
    - FDA orphan status for DMD & CF
  - gene replacement
  - stem cells - replace dystrophin

FYI - Click for more information on PTC 124
http://www.mda.org/research/070423dmd_ptc_124.html
Spinal Muscle Atrophy

Description
- An autosomal recessive hereditary disease characterized by progressive hypotonia (floppiness) and muscular weakness.

SMA major types:
- Type I (Werdnig-Hoffmann disease) - identified in patients from birth to age 6 months.
- Type II chronic infantile SMA - diagnosed in infants aged 6-12 months.
- Type III (Kugelberg-Welander disease) - diagnosed in children aged 2-15 years.

Epidemiology
- The most common degenerative neurologic disease in children
- The leading heritable cause of infant mortality
- Incidence - 1 case per 15,000-20,000
- 3-10 times more common in North Dakota
- Male:female = 2:1

Pathophysiology
- Absence of a neuron survival gene allows programmed cell death (apoptosis)
- Progressive degeneration of motor neurons from anterior horn cells in the spinal cord.

Manifestations
- Type I - shows at birth to 6 mo.
  - suspected with decreased fetal activity
  - floppy (hypotonic) newborn
  - unable to control head or roll over
  - tongue fasciculations - cardinal sign
Manifestations
- Type II - shows at 6-12 mo.
  ◆ weakness in lower extremities
  ◆ able to control head and sit up
  ◆ upper extremity tremors
  ◆ tongue fasciculations - cardinal sign

- Type III - shows at 2-15 yrs.
  ◆ tongue fasciculations - cardinal sign
  ◆ can walk early in life, then weaken
  ◆ wheelchair bound by 40 YO
  ◆ SMA does not affect cognition

Differential diagnosis
- Cerebral palsy
- Muscular dystrophy
- Myasthenia gravis
- Polymyositis
- Inflammatory polyneuropathy
- Infant botulism - honey

FYI - Click to download article on infant botulism

Diagnosis
- Clinical presentation
  ◆ progressive course of weakness
  ◆ tongue fasciculations

- Electrodiagnostics
  ◆ fasciculation potentials
  ◆ denervation

- Laboratory - prenatal DNA testing

Prognosis
- Type I SMA
  ◆ respiratory management has increased longevity
  ◆ 1 YO without ventilation
  ◆ >10 YO with noninvasive ventilation

- Type III SMA - may have normal life span

Management
- Supportive
  ◆ respiratory care
  ◆ nutrition
  ◆ physical therapy
  ◆ nursing
  ◆ end-of-life care - especially for parents
Critical Illness Polyneuropathy & Myopathy

**Description**
- First described in 1986
- Critical illness polyneuropathy (CIP) and critical illness myopathy (CIM) are complications of critical illness characterized by limb weakness and prolonged ventilator weaning.
- AKA - Intensive care unit-acquired weakness (debilitation)

**Epidemiology**
- Incidence - patient experiencing:
  - sepsis or systemic inflammation - 70%
  - multiple organ failure - 100%
  - ventilation for 4-7 D - 25-33%
  - ICU for 7 D - 49-77%

- Additional risk factors
  - corticosteroids
  - neuromuscular blocking agents
  - aminoglycosides; e.g., gentamycin
  - hyperglycemia
  - parenteral nutrition (TPN)

**Pathophysiology**
- Complex and unclear
- The neuromuscular system may be an additional organ in multiple organ failure (MOF)
- Muscle inactivity ==> atrophy

- Disruption of neuromuscular microcirculation by:
  - hyperglycemia
  - cytokines
- decreased anabolic hormones
- increased catabolic hormones
### Manifestations
- Muscle weakness, paralysis
- Absent deep-tendon reflexes
- Prolonged weaning from ventilator
- Longer duration of stay in ICU and hospital; therefore:
  - Increased costs ($66,000/patient)
  - Increased risk for morbidity; e.g., VAP

### Differential Diagnosis
- Myasthenia gravis
- Guillain-Barre’ syndrome
- Amyotrophic lateral sclerosis
- Polymyositis
- Multiple sclerosis
- Et cetera

### Diagnosis
- Primary aim is to rule out other diagnoses that may be treatable
- Clinical presentation
  - Generalized weakness
  - Weaning difficulty
- Electrodiagnostics - results may not justify the costs
- Laboratory - muscle biopsy to exclude myopathy

### Management
- Supportive
  - Respiratory care
  - Nutrition
  - Nursing
  - Physical therapy for recovery
- Intensive insulin therapy - maintain glucose 80-110 mg/dL
- Avoidance of potential triggers; e.g., steroids, neuromuscular blockers
- Mobilization in ICU

### Respiratory Assessment For Weakness
Respiratory muscle evaluation
- Important point - respiratory muscle weakness can be detected by bedside evaluations before any blood gas abnormality is present.

Physical signs
- rapid, shallow breathing - early sign
- accessory muscle recruitment
  - sternocleidomastoids
  - scalenes
  - external intercostals
  - abdominal - active expiration
- abdominal paradox - diaphragmatic fatigue

Blood gas analysis
- Oxygenation deficits - advanced disease
  - hypoventilation
  - ventilation-perfusion defects
    - atelectasis
    - pneumonia

Blood gas analysis
- Hypercapnia - advanced disease
  - weakness ==> rapid, shallow breathing
  - decreased sensitivity to CO2 (important)

Lung volumes and flows
- Lung volumes - restrictive pattern with initial normal compliance
  - decreased VC, TLC
  - normal FRC & RV
  - worsening with kyphoscoliosis - true restriction

Lung volumes and flows
- Comparison of erect, vs. supine VC
  - can be done at bedside
  - normal supine is 10% less than erect
  - greater difference ==> muscle weakness
### Lung volumes and flows
- Lung volumes are not sensitive measures of respiratory muscle strength.
- Lung volumes are compromised by reduced compliance; inspiratory pressure is not compromised - pressures are better indicators of strength.

### Expiratory flow rates
- Decreased FEV₁
- Normal FEV₁/FVC
- Decreased PEF ==> impaired cough effectiveness
- Peak cough flow >160 L/min - effective cough for adults

### Inspiratory/expiratory pressures
- **PI\textsubscript{MAX}**
  - Should be measured at FRC
  - Large range for normals
  - Sources of error:
    - Patient effort
    - Leaks
    - Technique - need to standardize

- **PE\textsubscript{MAX}**
  - Should be measured at TLC
  - Reflects coughing capability

FYI - Article with predicted MIP and MEP values

### Sniff nasal inspiratory pressure (SNIP)
- **Description**
  - Manometer connected via tube to one nostril
  - Other nostril remains open
  - Patient instructed to sniff maximally

- **Advantages**
  - Natural maneuver - easy to do
  - Reliability better than PI\textsubscript{MAX}
  - Validated for all age groups

- **Prognostic information** - SNIP < 40 cm H2O ==> nocturnal hypoxemia and risk for mortality (ALS patients)

- **Predicted values** - based on age and gender

FYI - Click to download article with predicted SNIP
Sleep studies (polysomnography)
- **Rationale**
  - to detect sleep-related hypventilation
  - determine need for nocturnal non-invasive ventilation

Sleep studies (polysomnography)
- **Baseline study - early in course of dx**
- **Follow-up**
  - appearance of signs of sleep apnea
  - daytime hypercapnia
  - nocturnal desaturation
  - periodically, to evaluate therapeutic regimen

**Respiratory Management**

**Quality of life and decisions**
- Noninvasive ventilation has increased survival time for NM patients
- Patients have their own perceptions on quality-of-life - likely to be better than we might think (must-read article for download below)

FYI - Click to download the must-read article on SMA

**Quality of life and decisions**
- Noninvasive ventilation has increased survival time for NM patients
- Patients have their own perceptions on quality-of-life - likely to be better than we might think
- Decisions about support should be made before any crisis
- Decisions should involve a healthcare team, patient and family

**Mechanical ventilation**
- Noninvasive ventilation indicated for:
  - sleep disordered breathing
  - hypercapnia
  - pulmonary infections
  - perioperative management
  - pregnancy
  - palliative, end-of-life care
Non-ventilatory care

Goal - delay need for ventilatory support

Problem

- ventilatory muscle weakness impairs cough
- secretion retention ==> recurrent pneumonia

Non-ventilatory care

Treatment strategies not used

- percussion and postural drainage - not routinely used
- bronchodilators - not indicated and may be harmful
- aerosolized mucolytics - ineffective
- deep suctioning

Non-ventilatory care

Treatment strategies

- manual cough assistance - tussive squeeze
- in/exsufflator device - indicated for MEP < 60 cm H2O
  - positive pressure for inflation
  - negative pressure for increased expiratory (cough) flow
  - usual pressures 40 to -40 cm H2O

Non-ventilatory care

Treatment strategies

- manual cough assistance - tussive squeeze
- in/exsufflator - indicated for MEP < 60 cm H2O
  - positive pressure for inflation
  - negative pressure for increased expiratory (cough) flow
  - usual pressures 40 to -40 cm H2O
  - may reverse atelectasis
  - improves symptoms and SPO2

Therapeutics

- in/exsufflator - cough assistance

Image Courtesy of Philips Respironics

Link to Emerson CoughAssist (TM) with video (2 min.)

Mechanical ventilation

Goals for ventilation

- increase survival time
- rest ventilatory muscles
- increase chemoreceptor sensitivity - decreases daytime PCO2
- improve sleep
Mechanical ventilation
- Negative pressure ventilation
  - as effective as NIPPV
  - cumbersome, difficult
- types:
  - iron lung
  - cuirass; e.g., Pneumo-Wrap
  - Hayek™ RTX (cuirass)

Click to see video of Hayek cuirass ventilator (5.5 min)
http://www.unitedhayek.com/presentations/movies/id/1

Mechanical ventilation
- Noninvasive positive-pressure ventilation (BiPAP)
  - treatment of choice for NM disease
  - progression - nocturnal to intermittent daytime use
  - mouthpiece during daytime
  - limitation - pressure injury from masks with continuous use

FYI - Link to RC article on NIPPV for pediatric patients
http://www.rcjournal.com/contents/08.06/08.06.0885.pdf

Mechanical ventilation
- PPV with tracheostomy
  - decision to continue support
  - required when:
    - risk for aspiration increases
    - NIPPV becomes ineffective
    - patient develops intolerance or injury from face mask

Mechanical ventilation
- Continued airway and ventilatory care
- Support for communication
- Education for home care
- End-of-life support

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