Patient Management

Pediatric Patients with Neuromuscular Disease

Teera Kijmassuwan, MD
Phetcharat Netmuy, B.N.S., MA

Oranee Sanmaneechai, MD : Preceptor
Thai boy 1 year old

Present with

- Respiratory failure at 6 month old
- Failure extubation 3 times with alveolar hypoventilation with apnea when he asleep
- Hypotonia with weakness
- Admit at Siriraj hospital for 6 month for treatment and work up
Differential diagnosis:

- Congenital central hypoventilation syndrome (CCHS)
- Neuromuscular disorder with respiratory failure
  - Congenital myopathies
  - Spinal muscular atrophy
Management

Respiratory care

- Tracheostomy
- Home ventilator

GI care

- Gastrostomy
- Feeding via kangaroo
Neuromuscular disease

- Heterogeneous group
  - Inherited
  - Acquired
- Progress muscle weakness and wasting
- Involvement of some component of the motor unit
- Prevalence of ~ 1 : 3000
Neuromuscular disease

eg. spinal muscular atrophy

eg. Charcot-Marie-Tooth disease [CMT]
Guillain Barrè Syndrome [GBS]/acute inflammatory demyelinating polyradiculoneuropathy [AIDP]
Chronic inflammatory demyelinating polyradiculoneuropathy [CIDP]

eg. myasthenia gravis [MG]
congenital myasthenic syndromes
Other organ complications

- Cardiac
- Respiratory
- Gastrointestinal
- Autonomic dysfunction
- Orthopedic
<table>
<thead>
<tr>
<th>Diagnosis</th>
<th>Age of Onset of Cardiac Disease</th>
<th>Cardiac Symptoms</th>
<th>Electrocardiographic Findings</th>
<th>Echocardiographic Findings</th>
<th>Prognosis</th>
</tr>
</thead>
</table>
| DMD                | Preclinical disease presents at 6‒8 years  
Clinical disease presents at 10‒18 years                                              | Asymptomatic CHF | Tall R waves in V1, V2  
Q waves  
BBB  
PACs, PVCs                                      | DCM  
RWMA  
MR                      | Death of cardiac or respiratory complications in the 20s to 30s                                     |
| BMD                | Preclinical disease presents at 10 to 20s  
Clinical disease rare in children, progresses with age, prevalent after 40 years    | Asymptomatic CHF | Q waves  
Tall R waves  
BBB                                      | DCM  
MR/MVP  
RV dilation  
RWMA                      | Heart failure in the 40s  
Poor prognosis once heart failure is present  
Heart transplant (eval) |
| DMD/BMD carrier    | Preclinical disease presents at 10 to 20s  
Clinical disease presents at 30 to 50s                                                   | Asymptomatic CHF | Tall R waves                                      | Normal DCM                       | Can have normal life expectancy  
Heart transplant (eval) in severe cases                                    |
| XL/DCM             | Clinical disease presents at 20 to 50s                                                      | Asymptomatic CHF | Q waves  
ST/T wave changes                              | DCM                                      | Poor survival after symptomatic  
Heart transplant (eval)                                                     |
| LGMD (types 1B, 2C, 2D, 2E, 2F, 2I) | Clinical disease mild and varies with diagnosis                                           | Asymptomatic CHF | Tall R waves  
Q waves  
1°, 2°, 3° AVB                                      | Normal DCM  
RWMA                      | Variable, depending on diagnosis  
Heart transplant (eval)                                                     |
| Myotonic dystrophy | Clinical disease presents in adulthood                                                      | Asymptomatic Syncope  
Sudden death          | 1°, 2°, 3° AVB  
AfiB/Aflutter  
VT/VF                                      | Normal DCM rare                | Progressive cardiac involvement, death in 50s  
Sudden death                                                                |
| Pompe disease      | Clinical symptoms have infantile onset                                                      | CHF               | Marked T wave  
Short PR interval                               | HCM                                      | Death usually occurs by 1 year                                             |
| Mitochondrial disorders | Variable onset, depending on diagnosis                                    | CHF  
Syncope             | ST/T wave abnormalities  
1°, 2°, 3° AVB  
Arrhythmias                                      | DCM  
HCM                      | Variable, depending on diagnosis                                 |
| Congenital myopathies | Variable onset, depending on diagnosis                                                    | Asymptomatic CHF | Tall R wave  
Q waves                                           | DCM                                      | Usually more dependent on skeletal and respiratory function  
Heart transplant (eval)                                                     |
Gastrointestinal complication

- Enteric nervous system
- Central nervous systems
- Autonomic nervous systems

**Box 4-1 Gastrointestinal Manifestations of Neuromuscular Diseases**

- Dysphagia
- Dyspepsia
- Gastroparesis
- Chronic intestinal pseudo-obstruction
- Bacterial overgrowth
- Weight loss
- Constipation
- Incontinence
Figure 5-1 Major pathways of the autonomic nervous system, with sympathetic innervation shown in red and parasympathetic innervation shown in blue. Preganglionic fibers are shown as solid lines, and postganglionic fibers are represented by dotted lines. GI, gastrointestinal.
Orthopedic

- Scoliosis
- Contracture
- Fractures
Management of Pediatric Patients with Neuromuscular Disease

Management

- Investigation
- Treatment
  - Specific treatment
  - Supportive treatment
Table 599-1  DISTINGUISHING FEATURES OF DISORDERS OF THE MOTOR SYSTEM

<table>
<thead>
<tr>
<th>LOCUS OF LESION</th>
<th>FACE</th>
<th>WEAKNESS</th>
<th>DEEP TENDON REFLEXES</th>
<th>ELECTROMYOGRAPHY</th>
<th>MUSCLE BIOPSY</th>
<th>OTHER</th>
</tr>
</thead>
<tbody>
<tr>
<td>Central</td>
<td>0</td>
<td>+</td>
<td>-</td>
<td>Normal or ↑</td>
<td>Normal</td>
<td>Seizures, hemiparesis, and delayed development</td>
</tr>
<tr>
<td>Ventral horn cell</td>
<td>Late</td>
<td>++++</td>
<td>++++</td>
<td>&gt; or =</td>
<td>0</td>
<td>Fasciculations and fibrillations</td>
</tr>
<tr>
<td>Peripheral nerve</td>
<td>0</td>
<td>+++</td>
<td>+++</td>
<td>&lt;</td>
<td>↓</td>
<td>Fibrillations</td>
</tr>
<tr>
<td>Neuromuscular junction</td>
<td>+++</td>
<td>+++</td>
<td>+++</td>
<td>=</td>
<td>Normal</td>
<td>Normal</td>
</tr>
<tr>
<td>Muscle</td>
<td>Variable (+ to ++++)</td>
<td>++</td>
<td>+</td>
<td>&gt;</td>
<td>↓</td>
<td>Short duration, small-amplitude motor unit potentials and myopathic polyphasic potentials</td>
</tr>
</tbody>
</table>

+ to ++++, varying degrees of severity; BSAP, brief duration, small amplitude, overly abundant motor unit potentials.

*Can also show unique features, such as in central core disease, nemaline myopathy, myotubular myopathy, and congenital fiber type disproportion.

Treatment of Motor Weakness

- **Pompe Disease**: Enzyme-replacement therapy
- **AIDP/GBS**: intravenous immunoglobulin (IVIg)
- **Dermatomyositis**: corticosteroids, second-line therapy for steroid-resistant
- **Duchenne muscular dystrophy (DMD)**: corticosteroids
- **MG**: anticholinesterase medications
Need with multisystemic involvement

Interdisciplinary care
  - Collaborative evaluation
  - Joint development of an individualized care plan for each patient

Table 2 Multidisciplinary/Interdisciplinary Health Care Team for Pediatric Neuromuscular Patients

<table>
<thead>
<tr>
<th>Anesthesia</th>
</tr>
</thead>
<tbody>
<tr>
<td>Cardiology</td>
</tr>
<tr>
<td>Critical care medicine</td>
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<tr>
<td>Endocrinology</td>
</tr>
<tr>
<td>Genetics</td>
</tr>
<tr>
<td>Nutrition</td>
</tr>
<tr>
<td>Orthopedics</td>
</tr>
<tr>
<td>Orthotics</td>
</tr>
<tr>
<td>Palliative care</td>
</tr>
<tr>
<td>Pediatrics (primary care)</td>
</tr>
<tr>
<td>Pediatric neurology</td>
</tr>
<tr>
<td>Pediatric surgery</td>
</tr>
<tr>
<td>Psychiatry</td>
</tr>
<tr>
<td>Psychology/neuropsychology</td>
</tr>
<tr>
<td>Pulmonary medicine</td>
</tr>
<tr>
<td>Social services</td>
</tr>
<tr>
<td>Support groups</td>
</tr>
<tr>
<td>Therapy: physical, occupational, speech</td>
</tr>
</tbody>
</table>
Goal: To optimize survival and quality of life

Care of the child with weakness

- Comorbid complications specific to each neuromuscular disease
- Management of motor dysfunction because of weakness
- Orthopedic complications of scoliosis and contractures
Early surveillance: an ECG and cardiac imaging (echocardiogram or MRI) at the time of diagnosis of muscular dystrophy is necessary

- Enable the initiation of heart failure therapy to prevent progressive deterioration of cardiac function
- Improve prognosis in patients with asymptomatic left ventricular dysfunction
Gastrointestinal management

- Excessive oral secretions
  - Glycopyrrolate
  - Botulinum toxin
- Feeding and swallowing difficulties
- Gastro-oesophageal reflux
  - Medical management
  - Surgical management
Orthopedic complication

- Rehabilitation
  - Muscle strength and function
  - Orthosis
- Orthopedic
  - Surgically treat scoliosis
  - Fractures
British Thoracic Society guideline for respiratory management of children with neuromuscular weakness

Jeremy Hull,¹ Roona Aniapravan,² Elaine Chan,³ Michelle Chatwin,⁴ Julian Forton,⁵ Jayne Gallagher,¹ Neil Gibson,⁶ Jill Gordon,⁷ Imelda Hughes,³ Renee McCulloch,⁸ Robert Ross Russell,² Anita Simonds⁴
Respiratory management

- Respiratory muscles are rarely spared
- Likelihood of respiratory impairment varies greatly among the different conditions
- Acute respiratory failure associated with respiratory infections are a frequent cause of hospitalization
- Chronic respiratory failure (CRF) is the most frequent cause of death
FIGURE 1
Components of neuromuscular respiratory failure.

Children with neuromuscular weakness should be evaluated by pediatric pulmonologist early in course of disease.

Clinical assessment of respiratory health should be part of every medical consultation:
- Progressive muscle weakness
- Ability to cope with respiratory infection
- Aspiration
- Progression of scoliosis
- Sleep-disordered breathing
กลไกการเกิดภาวะแทรกซ้อน

1. ความสามารถในการหายใจเข้าลดลง
2. ความสามารถในการไอลดลง
3. ความสามารถในการกลืนลดลง
4. ทางเดินหายใจส่วนต้นดีบแคบในขณะหลับ
Clinical finding

- **History**
  - Recurrent lower respiratory tract infection
  - Aspiration
  - Snoring
  - Sleep disturbance

- **Physical examination**
  - Chest wall deformity: scoliosis, pectus excavatum
  - Shortness of breathing: restrictive
Arterial blood gas

- SpO$_2$ < 94% at daytime
- PaCO$_2$ > 45 mmHg

Nocturnal hypoxemia / hypoventilation
Evaluation of chest function

- Vital capacity should be measured in all patients who are capable of performing spirometry
  - Generally achievable from 6 years of age
  - Inspiratory muscle strength and lung and chest wall compliance
Pulmonary function test
Pulmonary function test

- Neuromuscular disease
  - Restrictive pattern
  - Decrease Vital capacity and total lung capacity, functional residual capacity
  - Normal FEV\textsubscript{1} : FVC > 80%

- FVC : evaluate risk for nocturnal hypoventilation
  - > 60% predicted → Low risk
  - < 40% predicted → High risk
Pulmonary function test

- **FEV<sub>1</sub>**
  - < 40% predicted ➔ Sleep-associated respiratory disorder
  - < 20% predicted ➔ Daytime CO<sub>2</sub> retention

- **Maximum expiratory pressure (MEP)**
  - > 60 cmH<sub>2</sub>O : Adequate cough
  - < 45 cmH<sub>2</sub>O : Inadequate cough
Pulmonary function test

- Maximum inspiratory pressure (MIP)
  - < 60 cmH\(_2\)O: Need nocturnal assisted ventilation

- Inspiratory vital capacity (IVC) & MIP

<table>
<thead>
<tr>
<th>IVC</th>
<th>MIP (cmH(_2)O)</th>
<th>Hypoventilation</th>
</tr>
</thead>
<tbody>
<tr>
<td>&lt; 60% predicted</td>
<td>&lt; 45</td>
<td>Hypoventilation (REM)</td>
</tr>
<tr>
<td>&lt; 40% predicted</td>
<td>&lt; 40</td>
<td>Hypoventilation (all stage)</td>
</tr>
<tr>
<td>&lt; 25% predicted</td>
<td>&lt; 35</td>
<td>Diurnal respiratory failure</td>
</tr>
</tbody>
</table>
Cough peak flow

- Assessment of effective secretion clearance
- Over the age of 12 years
Evaluation of chest function

- Assessment for sleep-disordered breathing
  - Children with neuromuscular disease
    - Vital capacity of <60% predicted
    - Non-ambulant because of progressive muscle weakness
    - Who never attain the ability to walk
    - Symptoms of obstructive sleep apnea or hypoventilation
    - Clinically apparent diaphragmatic weakness
    - Rigid spine syndromes
  - Infants with weakness
Sleep-disorder evaluation

- Overnight pulse oximetry
- All children with abnormal overnight oximetry should undergo more detailed sleep monitoring with at least oxycapnography
How to monitor chest function?

- When there is doubt about the cause of sleep-disordered breathing
  - Overnight polysomnography
- Portable overnight oxycapnography or polygraphy in the home may be the most appropriate option for some patients
Management of respiratory problem

- Airway clearance and respiratory muscle training
  - Augmented cough techniques
  - Manual cough assist and air-stacking methods
  - Mechanical insufflation/exsufflation (MI-E)
  - Oscillatory techniques
    - High-frequency chest wall oscillation
    - Intrapulmonary percussive ventilation
  - Nebulised normal saline
  - Humidification
  - Use of NIV during airway clearance sessions can help prevent respiratory muscle fatigue
Assisted ventilation

- Treat symptom of nocturnal hypoventilation
- Treat symptom of daytime hypoventilation
- Reduce the frequency of hospital admission for chest infection
- Prevent chest wall deformity in young children
- Prolong life
  - NIV should be the first-line treatment
  - Nocturnal mechanical ventilation
  - Diurnal mechanical ventilation
Indication

- American Thoracic Society & European Consortium on Chronic Respiratory Insufficiency
- Indication for nocturnal ventilatory support
  1. Daytime hypercapnia (PCO₂ > 50 mmHg)
  2. Sleep hypercapnia (PCO₂ > 50 mmHg) + SpO₂ < 92%
  3. Recurrent pneumonia or Atelectasis

Panitch HB. Respiratory issues in management of children with neuromuscular disease. *Respir Care.* 2006;41:885-93
Ventilatory support

- Noninvasive
  - BiPAP

- Invasive
  - Ventilator
Contraindications to non-invasive ventilation

- Bulbar involvement and swallowing dysfunction
- Lack of patient motivation
- Inability to manage oropharyngeal secretion
- Mental status changes or cognitive impairment
- Cardiovascular instability
- History of pulmonary aspiration secondary to gastropharyngeal reflux and/or vocal cord paralysis
- Inability to fit or tolerate the mask interfaces
Indication for invasive pressure ventilation

- Failure to adequately ventilate with non-invasive ventilation
- Failure to tolerate non-invasive ventilation
- High level of dependence on assisted ventilation
Extubation criteria

- Presence of only minimal airway secretions
- Use of effective airway clearance methods (such as MI-E devices)
- Oxygen saturation more than 94% without supplemental oxygen for more than 12 h

Continuous NIV should be used immediately after extubation
NEUROMUSCULAR CARE IN NURSE ASPECT
Progress note

- D/C with home ventilator
- Plan weaning diurnal ventilator to nocturnal
- Change tracheostomy tube and gastrostomy tube every month
Thank you for your attention