Update on Respiratory Care in Myopathies

Anita Simonds
Royal Brompton Hospital

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General good health

• Sensible diet, posture, general fitness, *Don’t smoke*
• Prevention: Flu & pneumococcal vaccination, physiotherapy, swallowing assessment, reflux, scoliosis
• Prompt recognition of chest infections and plan eg. low threshold for antibiotics or reserve course
• Appropriate management of other common problems eg. asthma, anaemia
Breathing problems

- Recurrent infections: as above, prompt rotating antibiotics, non-invasive ventilation (NIV), cough assistance if cough peak flow reduced
- Chronic resp insufficiency: NIV for sleep disordered breathing, daytime resp failure, weaning. Combination with expiratory cough aids
- Invasive ventilation: profound swallowing dysfunction, unable to manage on non-invasive support
- Anticipatory negotiated care planning: when to do what
Key Points

• Identify children and adults *at risk* of breathing problems eg. childhood nemaline myopathy, multicore myopathy, X linked myotubular myopathy
• Assess on an individual basis
• Anticipate and prevent breathing problems, rather than wait till they occur!
• Remember - breathing problems are *very* treatable
Clinical assessment

- Inspiratory, expiratory, bulbar muscle weakness
  - REM related sleep disordered breathing
    - FVC < 60% pred
  - Ineffective cough
    - Cough peak flow < 270l/min
  - NREM and REM sleep disordered breathing
    - FVC < 40% pred
  - Chest infections
  - Daytime ventilatory failure
    - FVC < 20% pred

Intervention

- Physical examination, pulmonary function, cough peak flow, resp muscle strength
- Chest radiology, sleep study.
- Swallow function
- Intervention: cough assistance
- Non-invasive ventilation, combination with cough assist, PEG/PEJ, T-IPPV
Current Assessment guidelines

- Ventilatory support in Congenital Neuromuscular Disorders – congenital myopathies, congenital muscular dystrophies and SMA II Neuromusc Disord 2004;14:56-69
- MDC Recommendations for the Respiratory care of adults with Muscle Disorders (Neuromusc Disord 2006 in Press)
- ATS Consensus Statement: Respiratory Care of the patient with Duchenne Muscular Dystrophy AJRCCM;2004:456-65
Assessment of respiratory function

- Estimate probability of respiratory complications: diagnosis, age, co-morbidity, intercurrent events
- Assessment of respiratory symptoms & signs [D]
- Measurement of Vital capacity (VC) (including supine if practical) and SaO2 annually [D]
- Tests of inspiratory and expiratory muscle strength and cough peak flow may be helpful [D]
- Patients with VC < 1l (DMD) or with rapid decline of VC should be assessed more frequently
- Overnight monitoring annually if VC <60% or symptoms of nocturnal hypoventilation [D]
- Arterial blood gases should be measured if SaO2 <93% or symptoms of noct hypventilation
- Plus: scoliosis, nutritional, swallowing and cardiac assessment : ECG, 24 hour ECG, Echocardiogram
Patterns/evolution respiratory muscle weakness

Normal insp: exp strength ratio >1

ie: greater expiratory muscle involvement in SMA

M. Chatwin et al. 2004 ATS
What type of sleep study?

Nocturnal hypoventilation

Normal overnight oximetry (on air) makes significant SDB unlikely
Overnight monitoring
Transcutaneous CO2 and SaO2
Control of nocturnal hypoventilation with NIV

A) Spontaneous ventilation

B) On NIV
Age at start of NIV in NMD children

N=40 Type II

Diagnostic category

Age Distribution at start of NIV

Mean (yrs)
- SMA 5.7
- Myopathy 7.3
- CMD 11.6
- DMD 13.9

Eur Respir J
2000;16: 476-81
Mechanical In-Exsufflation in Children

- n = 62
  - Duchenne MD (16)
  - Congenital myopathy (12)
  - Non-specific NMD (12)
  - SMA Type I (8)
  - SMA Type II (14)
- For group, median age 12.6 yrs (range 3 mo to 28.6 yrs)

- No correlation of pressures with age or underlying disease
- Median insufflation pressure: +30 cm H₂O
  - range 15 - 40 cm H₂O
- Median exsufflation pressure: -30 cm H₂O
  - range -20 to -50 cm H₂O
- Daily to q 4hr (most BID)

When to initiate NIV in NMD?

- To *prevent* respiratory decompensation?
- To *treat* ventilatory failure
- To *treat* sleep disordered breathing?
- To *improve* chest wall & pulmonary development?
- To *treat/prevent* chest infections
- Anticipatory use: Surgery
  - Intermittent muscle weakness
  - Pregnancy
Randomised controlled trial of NIV in nocturnal hypoventilation in congenital neuromusculo-skeletal disease: trial design

n = 48
Total number of patients screened

Nocturnal hypoventilation
TcCO2 > 6.5 kPa

Daytime normocapnia
n = 26

Group 1 Randomised to control
No NIV
n = 12

Group 2 Randomised to NIV
n = 14

Daytime hypercapnia
n = 19

Group 3 Elective NIV
n = 19

Ward, Chatwin, Heather & Simonds Thorax 2005; 60:1019-24
Method

Gp 1 &2
Median age 18 yr Noct TcCO2 9.15 kPa
Diurnal PaCO2 5.9 kPa PaO2 10.5 kPa
DMD, CMD, SMA II, Beals syndr

A priori safety criteria for Gp 1
Daytime PaCO2 > 6.5
Worsening symptoms of nocturnal hypoventilation
Recurrent RTIs (>3/yr)
Failure to thrive
Acute ventilatory decompensation
**Group 1** Control
Randomised to follow-up
n=12

- 2 drop outs

Completed 24 mths
n=10

- Completed 24 months without NIV
  n=1

- Fulfilled criteria for NIV and failed F/U
  n=9

**Group 2**
Randomised to NIV
n=14

- 2 F/U elsewhere

Completed 24 mths
n=12

- Continued NIV
  n=9

- Elected not to receive NIV
  n=3

- Required emergency NIV
  n=2

**Group 3**
Elective NIV
n=19

- 2 drop outs

Completed 24 mths NIV
n=17

- Completed 24 mths

Results
### Results

**Summary of group 1 patients failing follow-up only**

<table>
<thead>
<tr>
<th>Patient ID</th>
<th>Months from entry onto study (0 – 24)</th>
<th>Reason for failure of follow-up only</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td></td>
<td>↑ daytime PaCO₂</td>
</tr>
<tr>
<td>2</td>
<td></td>
<td>↑ daytime PaCO₂</td>
</tr>
<tr>
<td>3</td>
<td></td>
<td>↑ TcCO₂ + ↓ PFTs</td>
</tr>
<tr>
<td>13</td>
<td></td>
<td>↑ daytime PaCO₂</td>
</tr>
<tr>
<td>14</td>
<td>NO NIV</td>
<td>↑ daytime PaCO₂</td>
</tr>
<tr>
<td>15</td>
<td></td>
<td>Symptomatic relief</td>
</tr>
<tr>
<td>16</td>
<td></td>
<td>Failure to thrive + acute pneumonia</td>
</tr>
<tr>
<td>17</td>
<td></td>
<td>Symptomatic relief</td>
</tr>
<tr>
<td>25</td>
<td></td>
<td></td>
</tr>
<tr>
<td>26</td>
<td></td>
<td>↑ daytime PaCO₂</td>
</tr>
</tbody>
</table>

### % of group receiving NIV

<table>
<thead>
<tr>
<th>% of group receiving NIV</th>
<th>Baseline</th>
<th>6 months</th>
<th>12 months</th>
<th>18 months</th>
<th>24 months</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>0%</td>
<td>50%</td>
<td>70%</td>
<td>70%</td>
<td>90%</td>
</tr>
</tbody>
</table>

i.e. 9/10 patients met criteria to receive NIV by end of study (70% within 1 year)

Ward et al Thorax 2005;60:1019-24
Results: Health status

SF 36: General health

<table>
<thead>
<tr>
<th>Group</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>Group 1</td>
<td>Controls no NIV</td>
</tr>
<tr>
<td>Group 2</td>
<td>Randomised to NIV</td>
</tr>
<tr>
<td>Group 3</td>
<td>Elective NIV</td>
</tr>
</tbody>
</table>

Inference: Neuromuscular patients with nocturnal hypoventilation are likely to progress to daytime hypercapnia within 12-24 months.
### Influence of noninvasive ventilation (NIV) on sleep-breathing and sleep

<table>
<thead>
<tr>
<th></th>
<th>Before NIV</th>
<th>During NIV</th>
<th>p-value</th>
</tr>
</thead>
<tbody>
<tr>
<td>RDI·h⁻¹</td>
<td>10.5±13.1</td>
<td>3.1±3.5</td>
<td>&lt;0.001</td>
</tr>
<tr>
<td>REM-RDI·h⁻¹</td>
<td>20.5±21.1</td>
<td>3.0±5.3</td>
<td>&lt;0.001</td>
</tr>
<tr>
<td>Arousal index·h⁻¹</td>
<td>20.6±14.3</td>
<td>10.2±3.8</td>
<td>&lt;0.001</td>
</tr>
<tr>
<td>Light-sleep %</td>
<td>55±12</td>
<td>44±13</td>
<td>&lt;0.05</td>
</tr>
<tr>
<td>Slow-wave-sleep %</td>
<td>24±9</td>
<td>34±9</td>
<td>&lt;0.05</td>
</tr>
<tr>
<td>REM-sleep %</td>
<td>18±6</td>
<td>20±6</td>
<td>0.18</td>
</tr>
</tbody>
</table>

RDI: respiratory disturbance index per hour sleep; REM-RDI: respiratory disturbance index per hour rapid eye movement (REM)-sleep; arousal index: electroencephalographic arousals per hour sleep.
Quality of life in Duchenne MD

No correlation between QoL and disability or need for NIV

Kohler et al. AJRCCM 2005;172:1032-6

Open circles/bars = spont breathing
Black circles/hatched = NIV users
Preventative use of home NIV to reduce hospital admissions in children with Type I & 2 SMA: Caregiver strain scores

Children aged 10 mths to 14 years with SMA Type 1.8-2.4

Chatwin M, Simonds AK
ATS 2006
Anticipatory Care Plan

- Identify high risk cases
- 6-12 mthly resp assessment: symptoms, signs, respiratory measurement – PFTs, cough PF, sleep studies
- Discussion of options for respiratory support and timing.
- Negotiated care plan
- Guidance and education for chronic care
- Planned introduction of NIV
- Cough and secretion management : stepped plan
- Immunizations, low threshold for antibiotics
- Regular nutrition and swallowing assessment
- Consideration PEG/PEJ if inadequate nutrition, swallowing
- Rapid access to specialty medical care providers
- Perioperative management plan & liaison with anaes/ICI/surgical team
- BE POSITIVE!!
Thank you

A.Simonds@rbht.nhs.uk