



Update on Respiratory Care in Myopathies

Anita Simonds

Royal Brompton Hospital

Myotubular Trust Family Conference Sept 8 2007

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

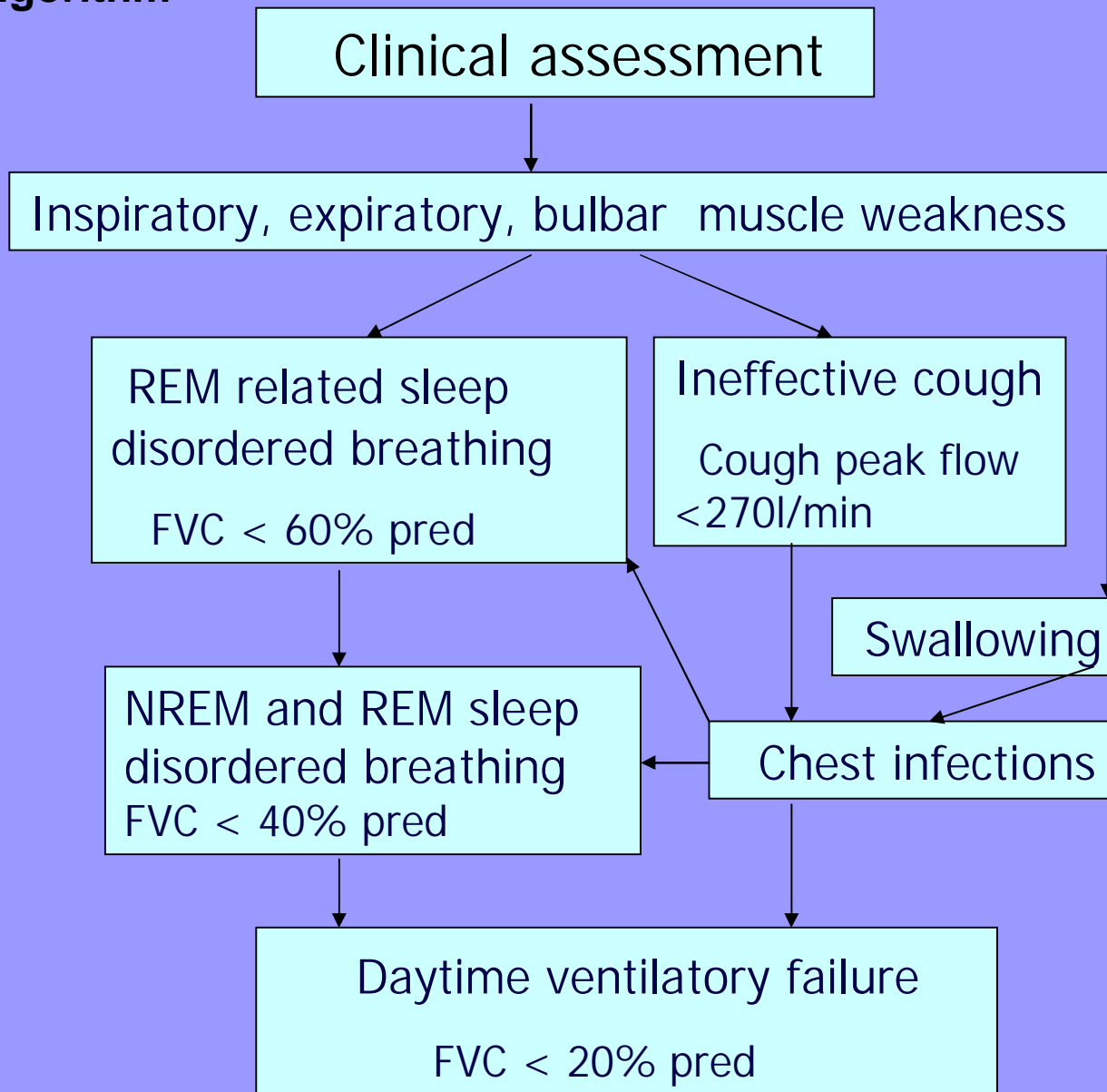
Breathing problems

- 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 algorithm



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

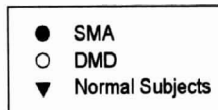
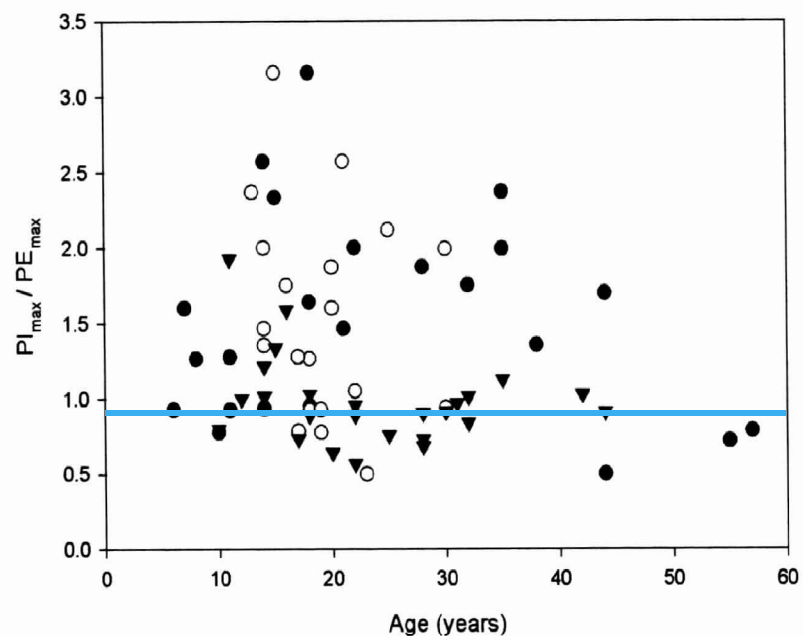
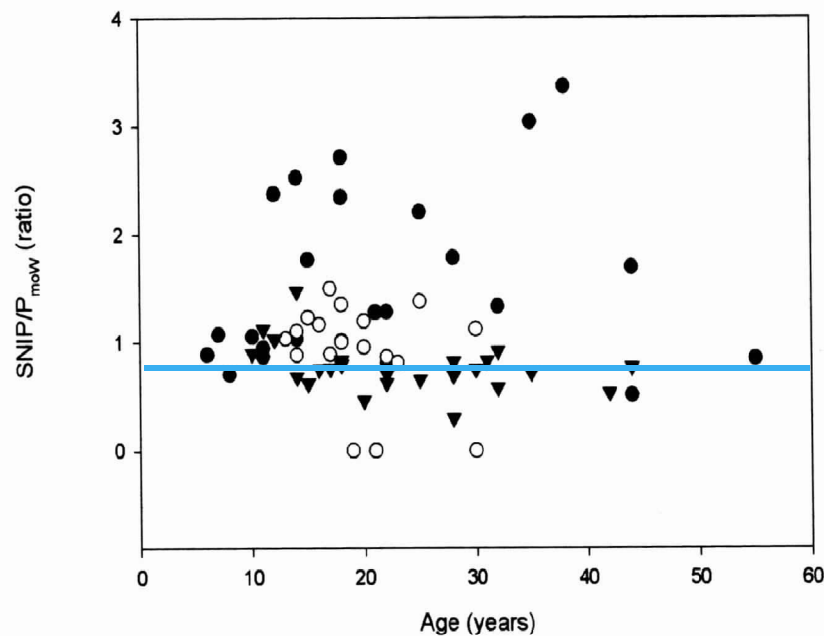
Current Assessment guidelines

- Ventilatory support in Congenital Neuromuscular Disorders – congenital myopathies, congenital muscular dystrophies and SMA II Neuromusc Disord 2004;14:56-69
- *Recommendation for Respiratory care of children with Spinal Muscular Atrophy Type II & III Neuromusc Disord 2003;13:184-89*
- *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
- Consensus Conference: Clin indications for NIV in CRF Chest 1999;116: 521-4

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 hypoventilation
- Plus: scoliosis, nutritional, swallowing and cardiac assessment : ECG, 24 hour ECG, Echocardiogram

Patterns/evolution respiratory muscle weakness

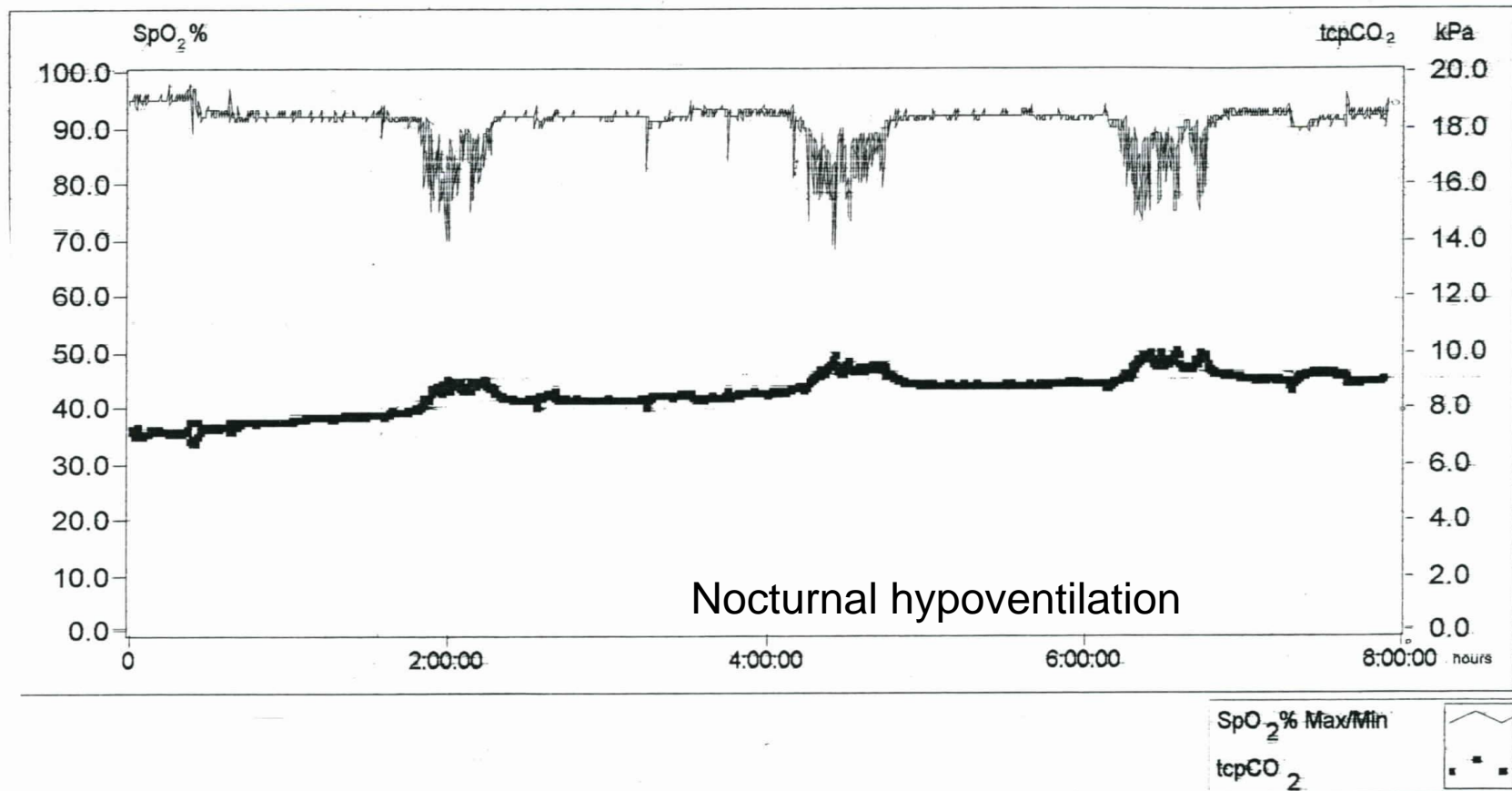


Normal insp: exp strength ratio >1

ie: greater expiratory muscle involvement in SMA

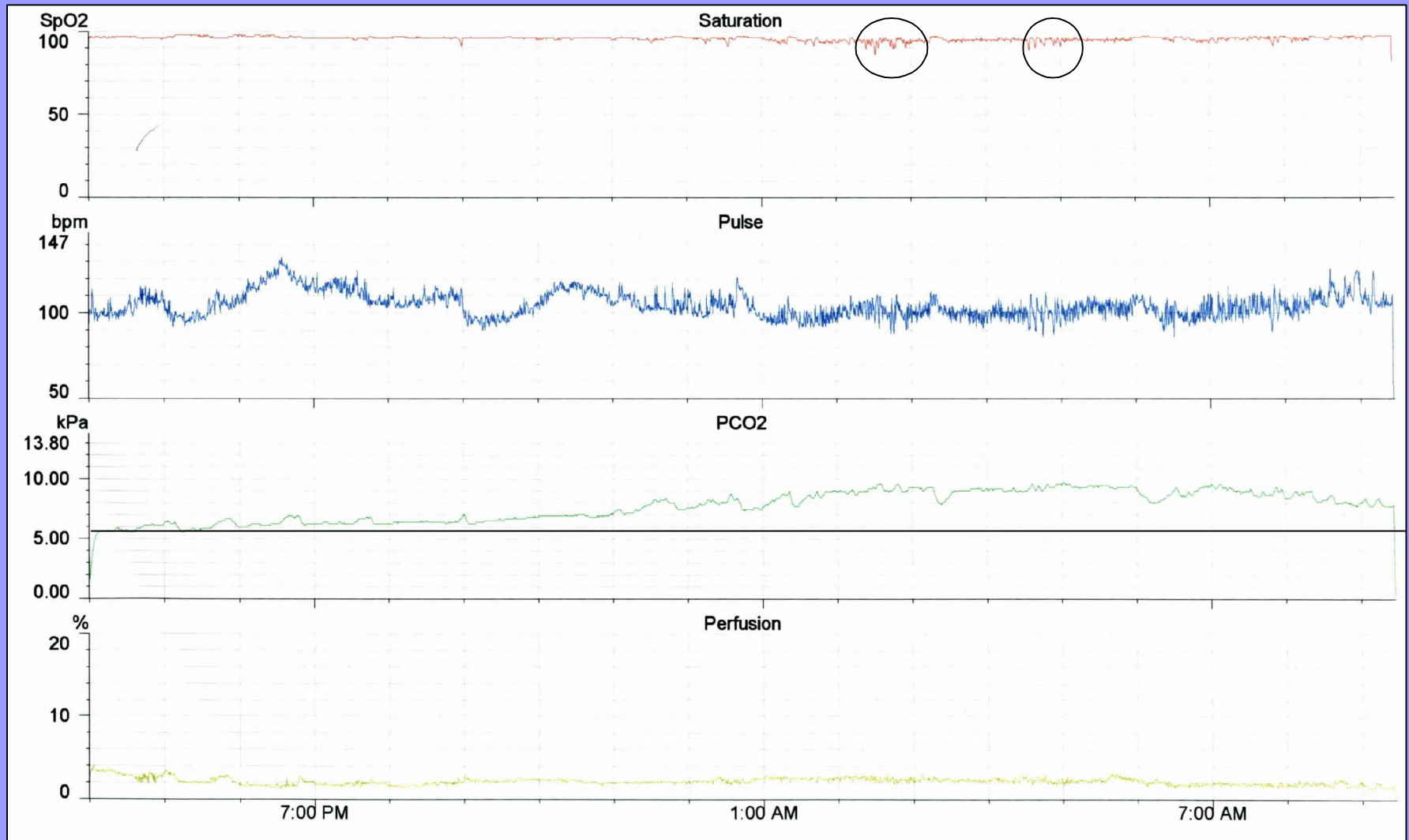
M.Chatwin et al 2004 ATS

What type of sleep study?

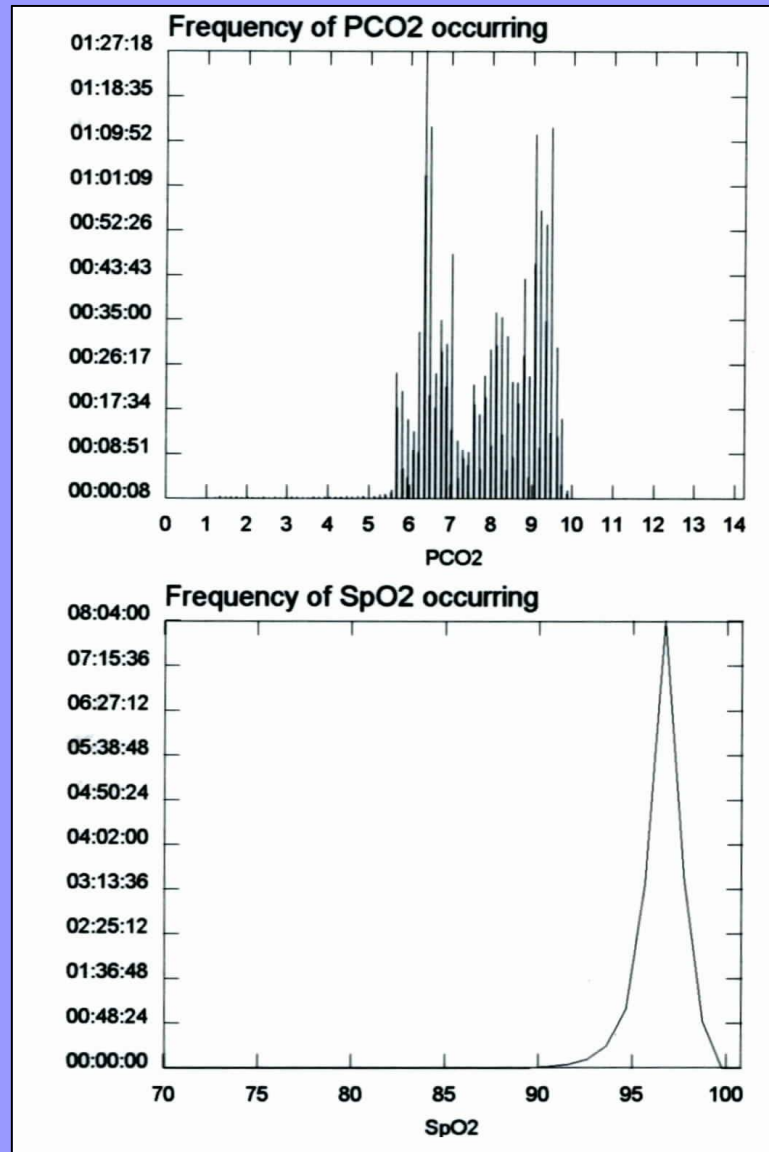


Normal overnight oximetry (on air) makes significant SDB unlikely

Overnight monitoring

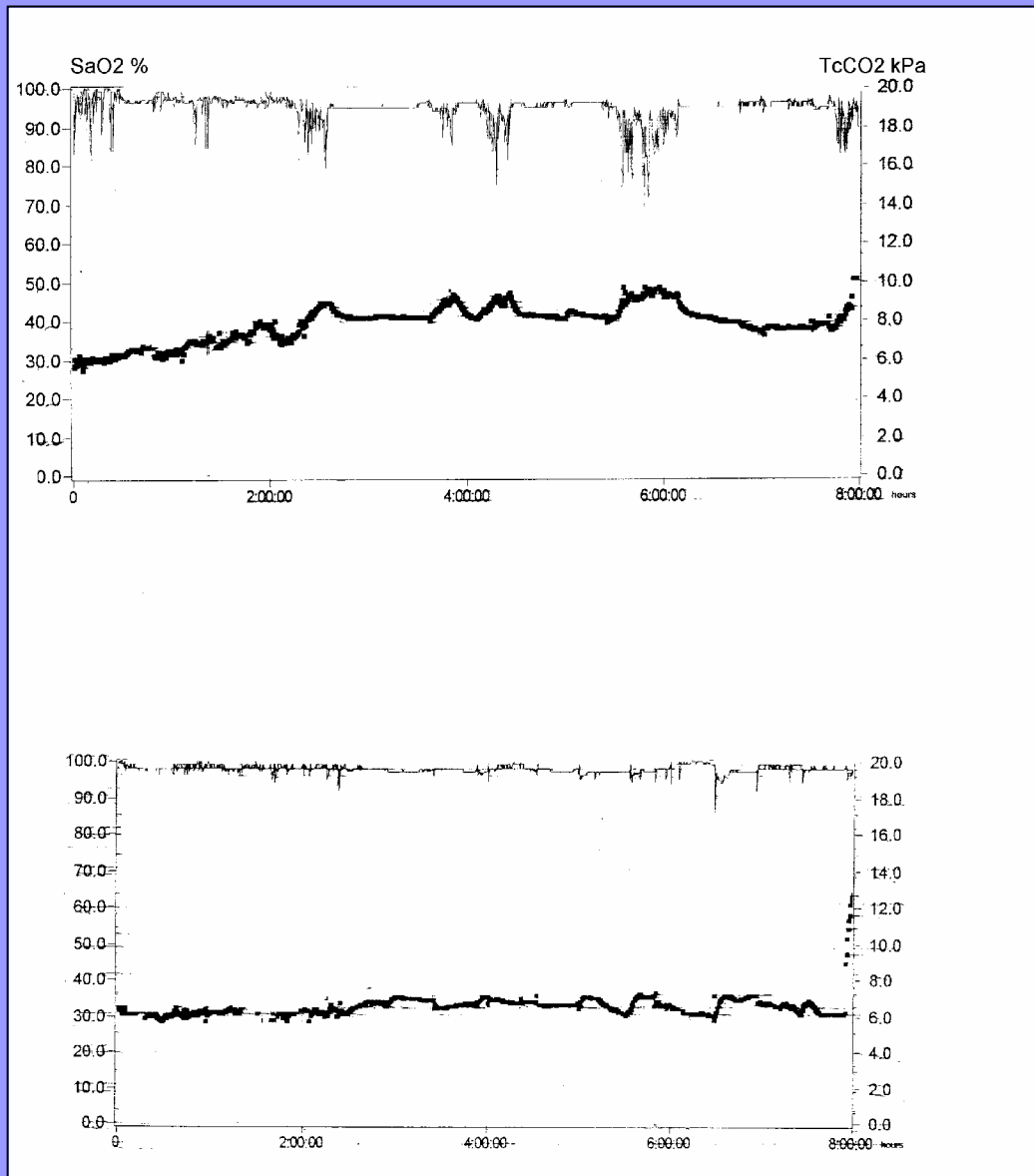


Transcutaneous CO₂ and SaO₂



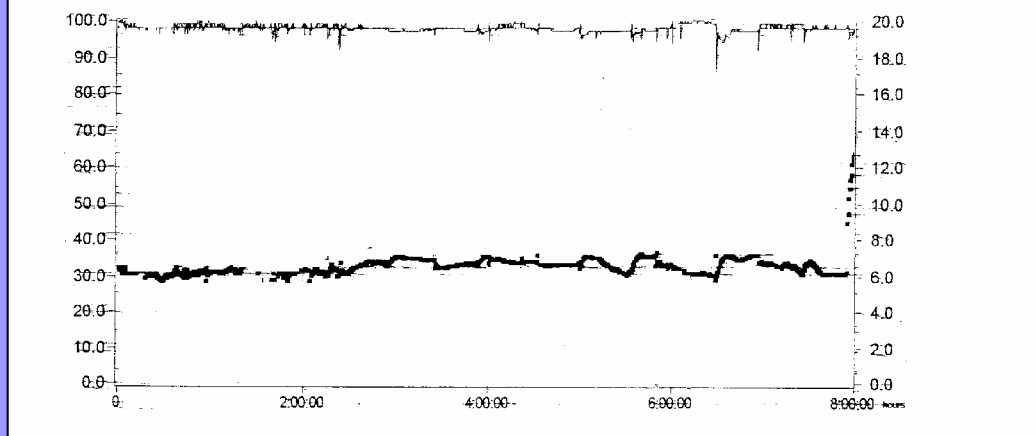
Control of nocturnal hypoventilation with NIV

A)



Spontaneous ventilation

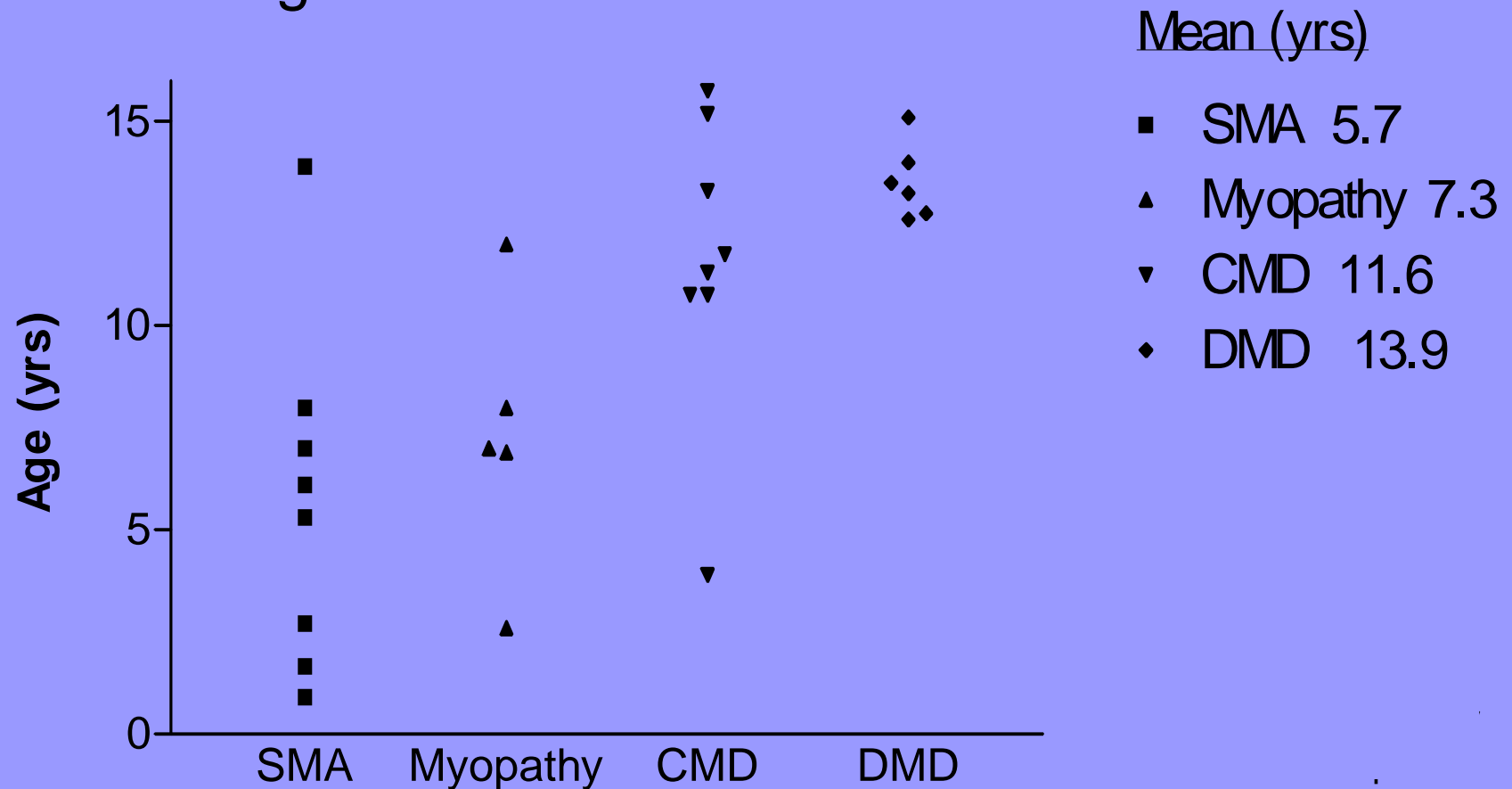
B)



On NIV

Age at start of NIV in NMD children

Age Distribution at start of NIV



N=40

Type II

Diagnostic category

Eur Respir J
2000;16: 476-81

Mechanical In-Exsufflation in Children

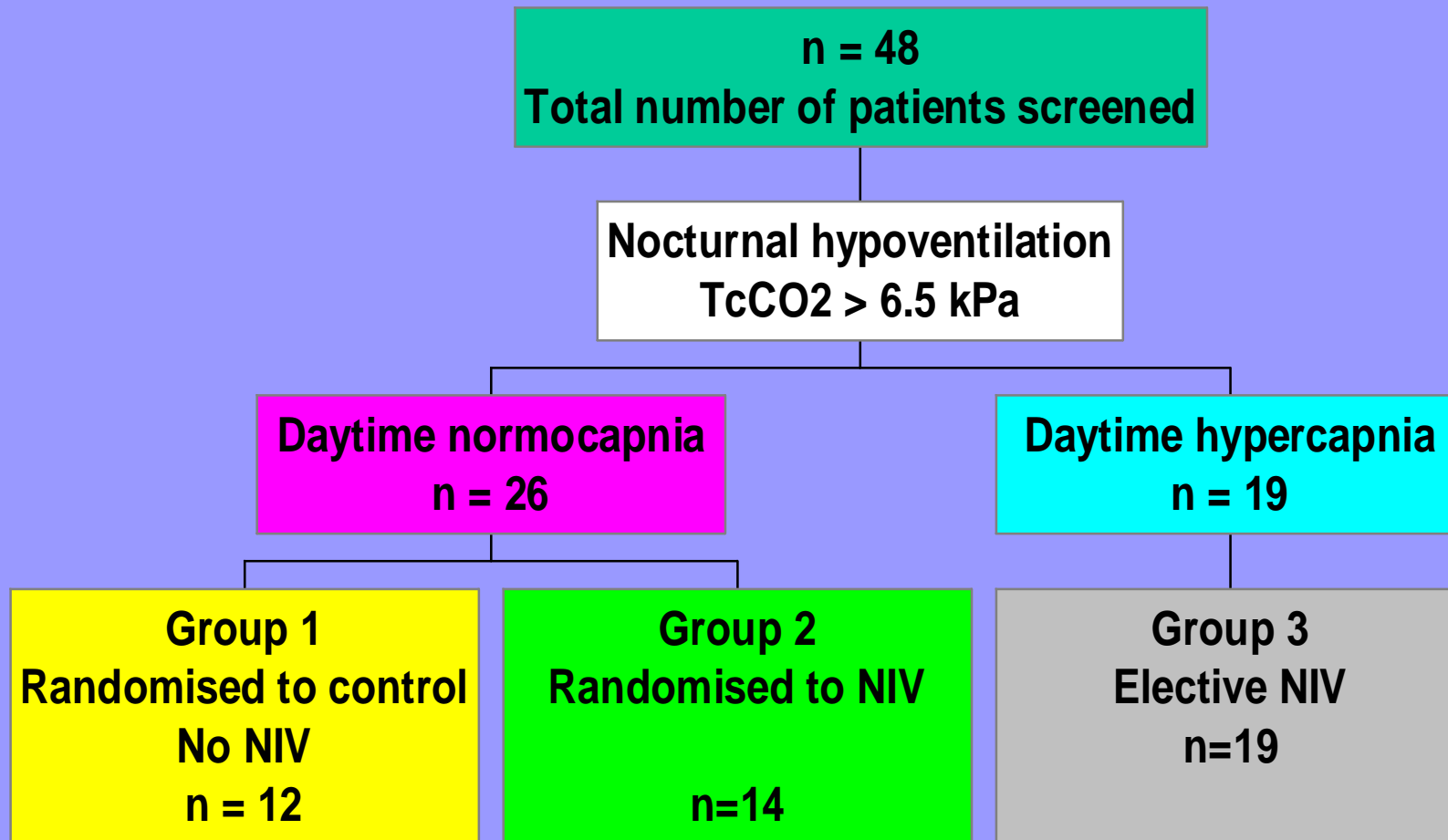
- n = 62
 - Duchenne MD (16)
 - Congenital myopathy (12)
 - Non-specific NMD (12)
 - SMA Type I (8)
 - SMA Type II (14)
- For group, median age 12.6 yrs (range 3 mo to 28.6 yrs)

- 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



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

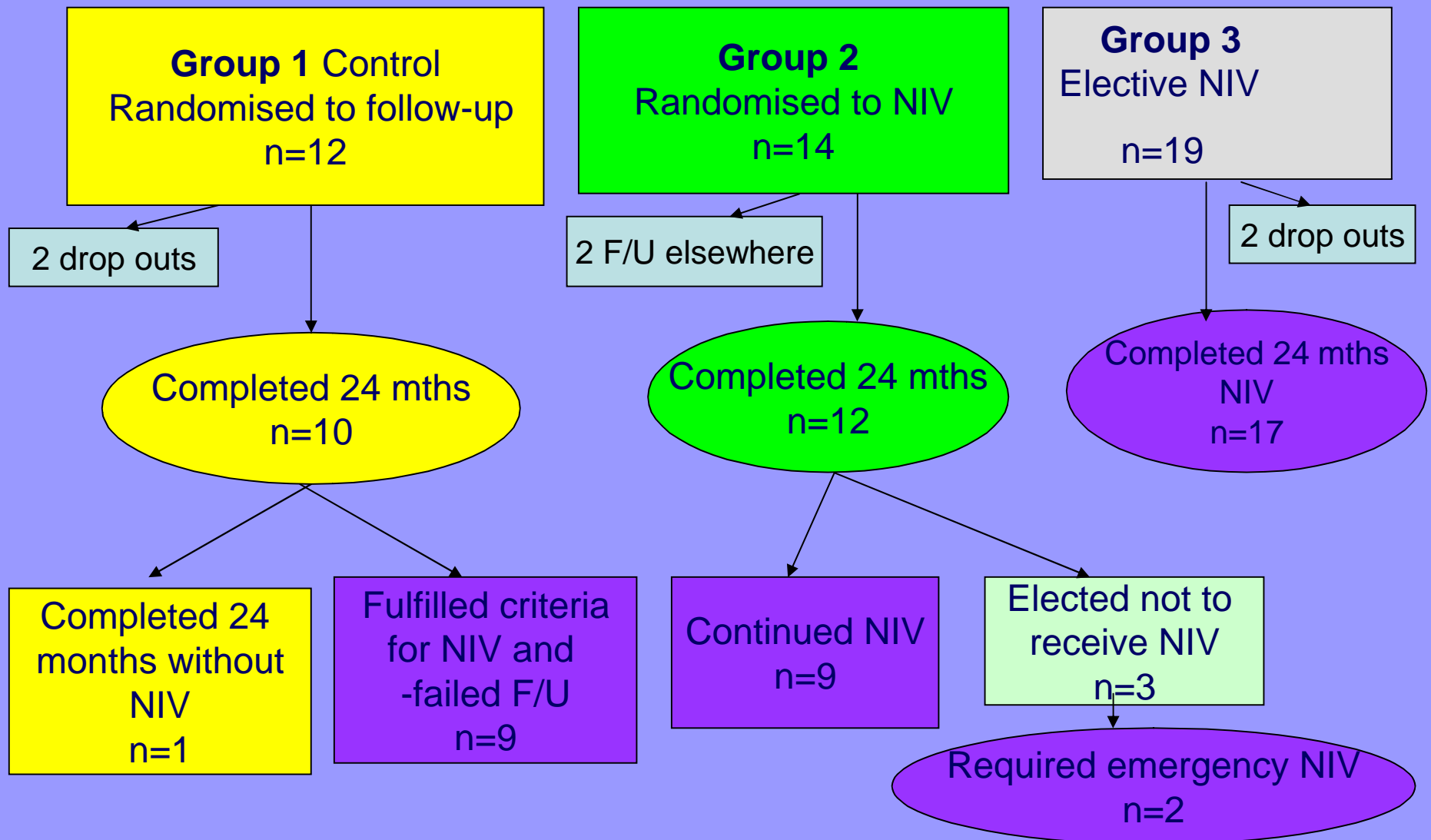
Worsening symptoms of nocturnal hypoventilation

Recurrent RTIs (>3/yr)

Failure to thrive

Acute ventilatory decompensation

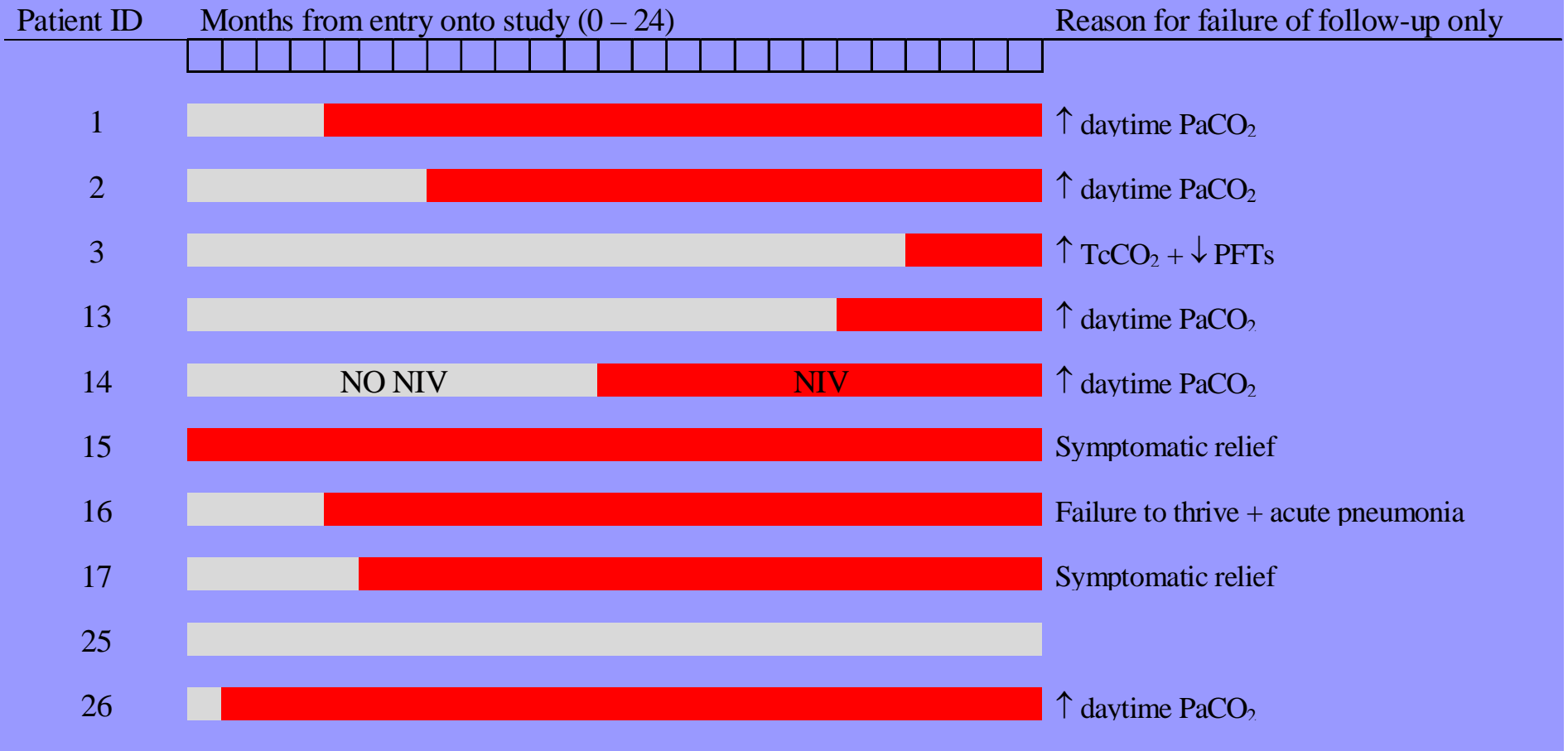
Results



Results

Ward et al Thorax 2005;60:1019-24

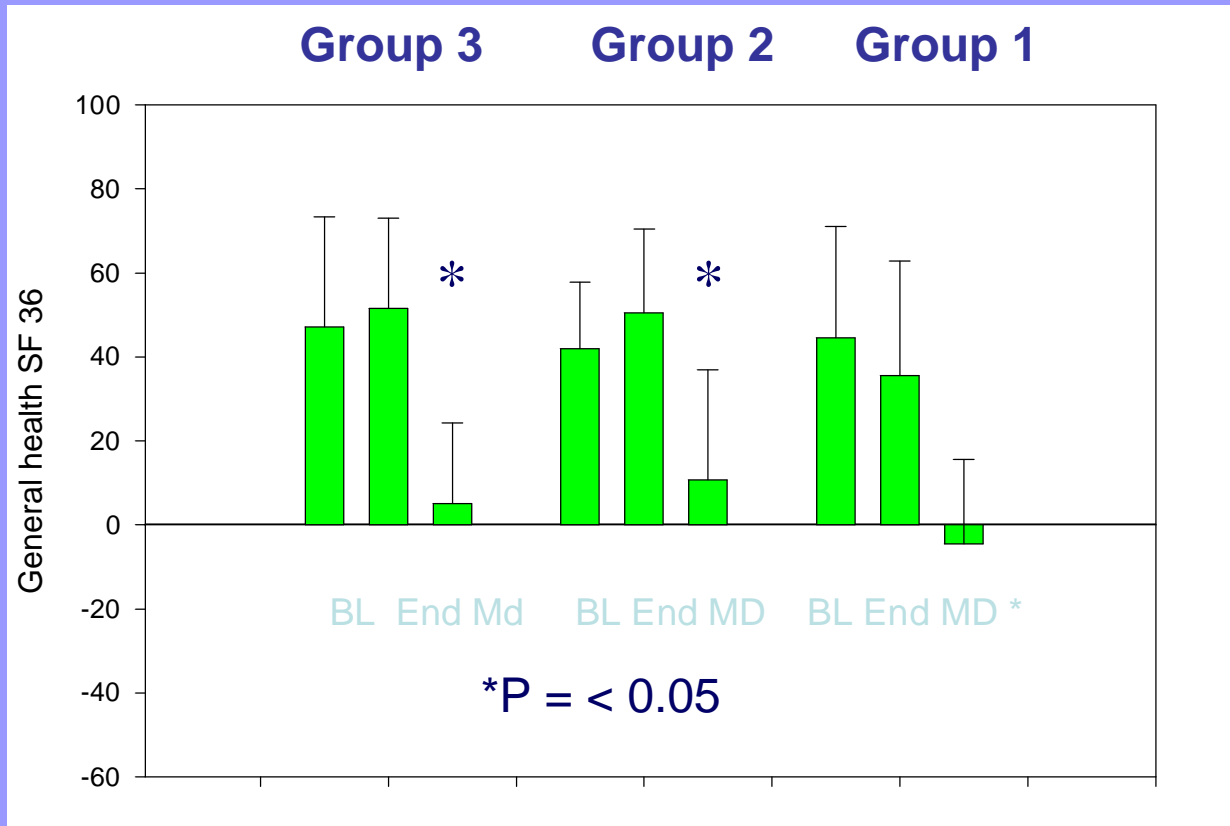
Summary of group 1 patients failing follow-up only



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

Results: Health status

SF 36 : General health



BL- Baseline score
End - End of trial score
MD – mean difference

Group 1 Controls no NIV **Group 2** Randomised to NIV **Group 3** Elective NIV

Inference: Neuromuscular patients with nocturnal hypoventilation are likely to progress to daytime hypercapnia within 12-24 months

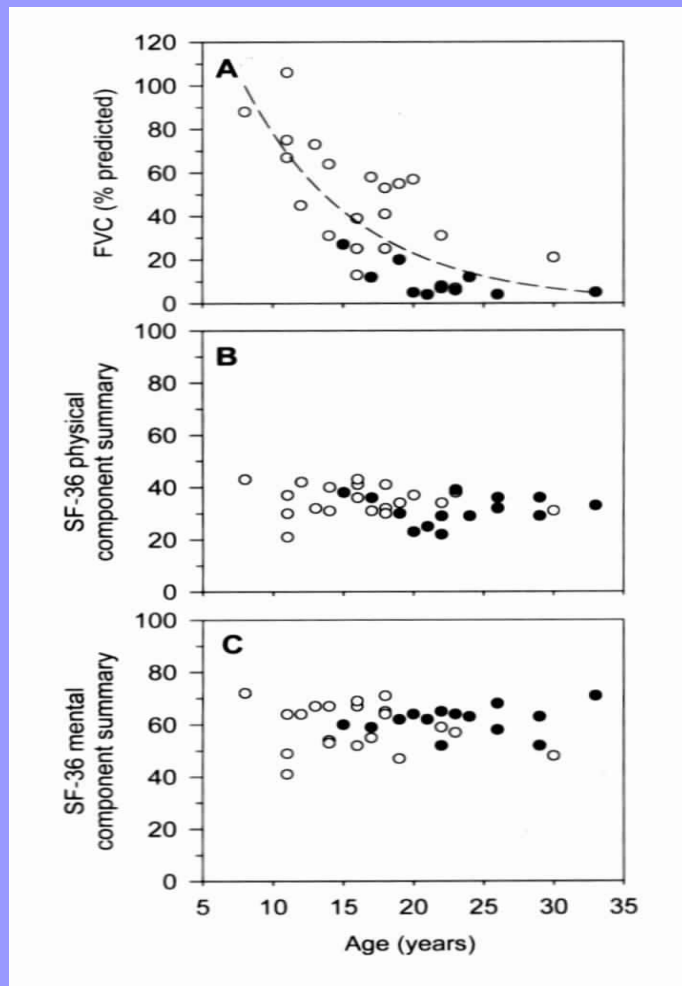
Effect of NIV on sleep quality

Influence of noninvasive ventilation (NIV) on sleep-breathing and sleep

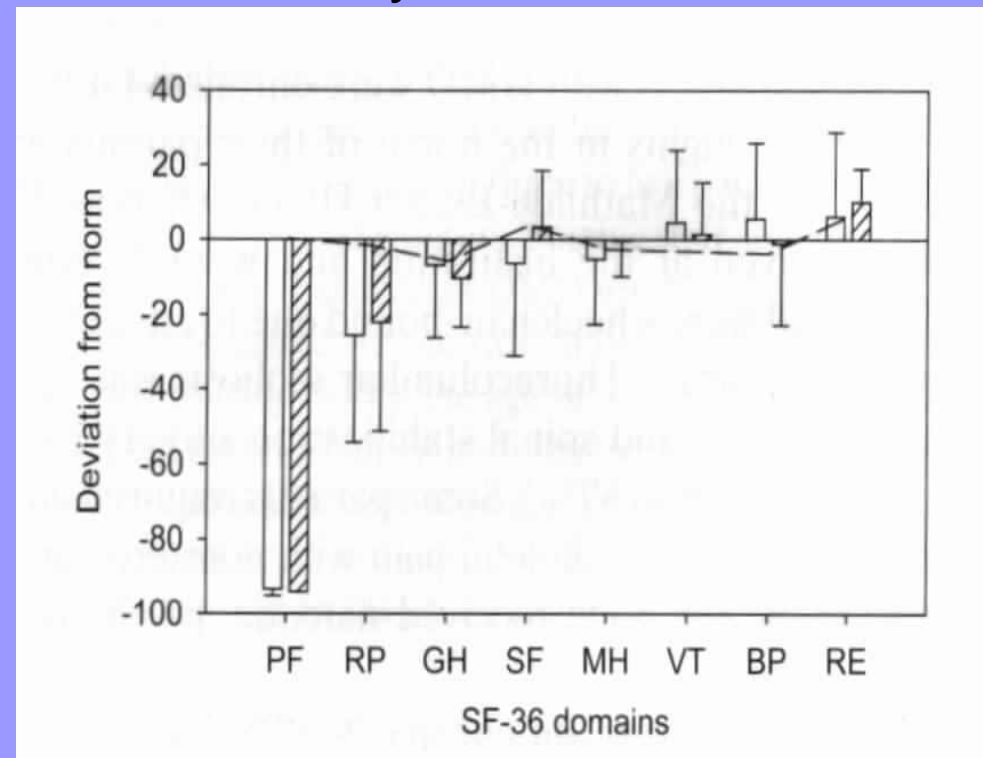
	Before NIV	During NIV	p-value
RDI·h ⁻¹	10.5±13.1	3.1±3.5	<0.001
REM-RDI·h ⁻¹	20.5±21.1	3.0±5.3	<0.001
Arousal index·h ⁻¹	20.6±14.3	10.2±3.8	<0.001
Light-sleep %	55±12	44±13	<0.05
Slow-wave-sleep %	24±9	34±9	<0.05
REM-sleep %	18±6	20±6	0.18

RDI: respiratory disturbance index per hour sleep; REM-RDI: respiratory disturbance index per hour rapid eye movement (REM)-sleep; arousal index: electrocephalographic arousals per hour sleep.

Quality of life in Duchenne MD



No correlation between QoL and disability or need for NIV

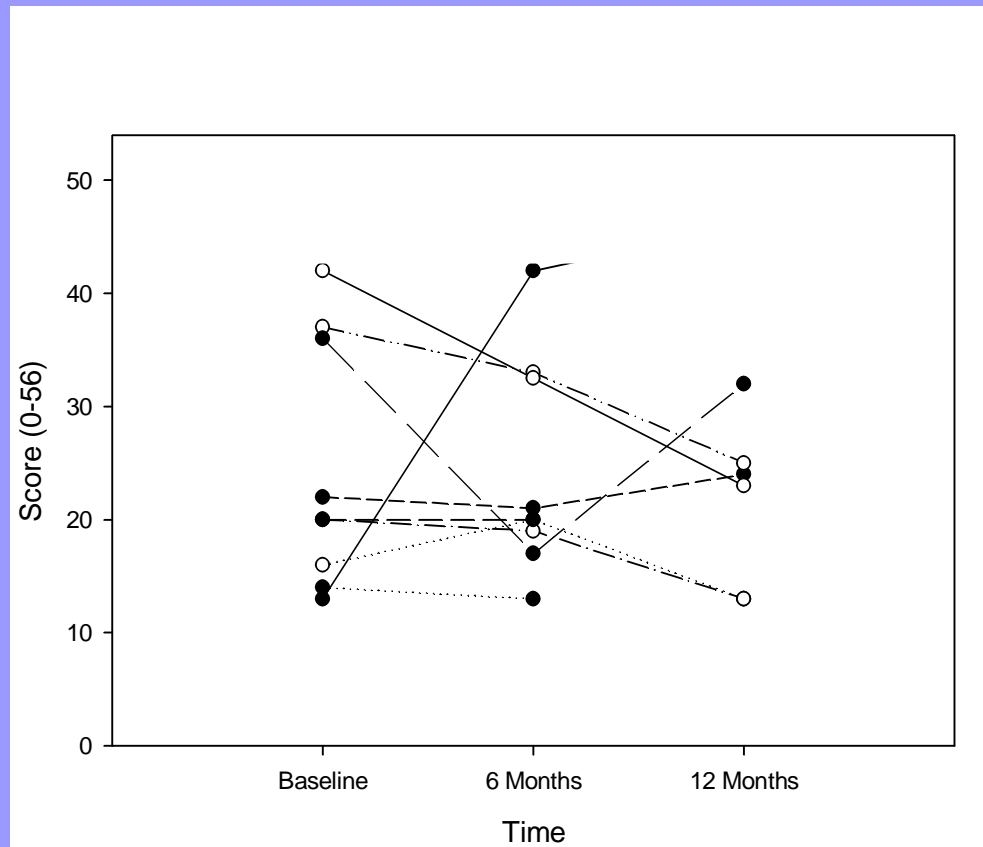


Kohler et al AJRCCM 2005;172:1032-6

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



Chatwin M, Simonds AK
ATS 2006

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

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