Respiratory management of SMA

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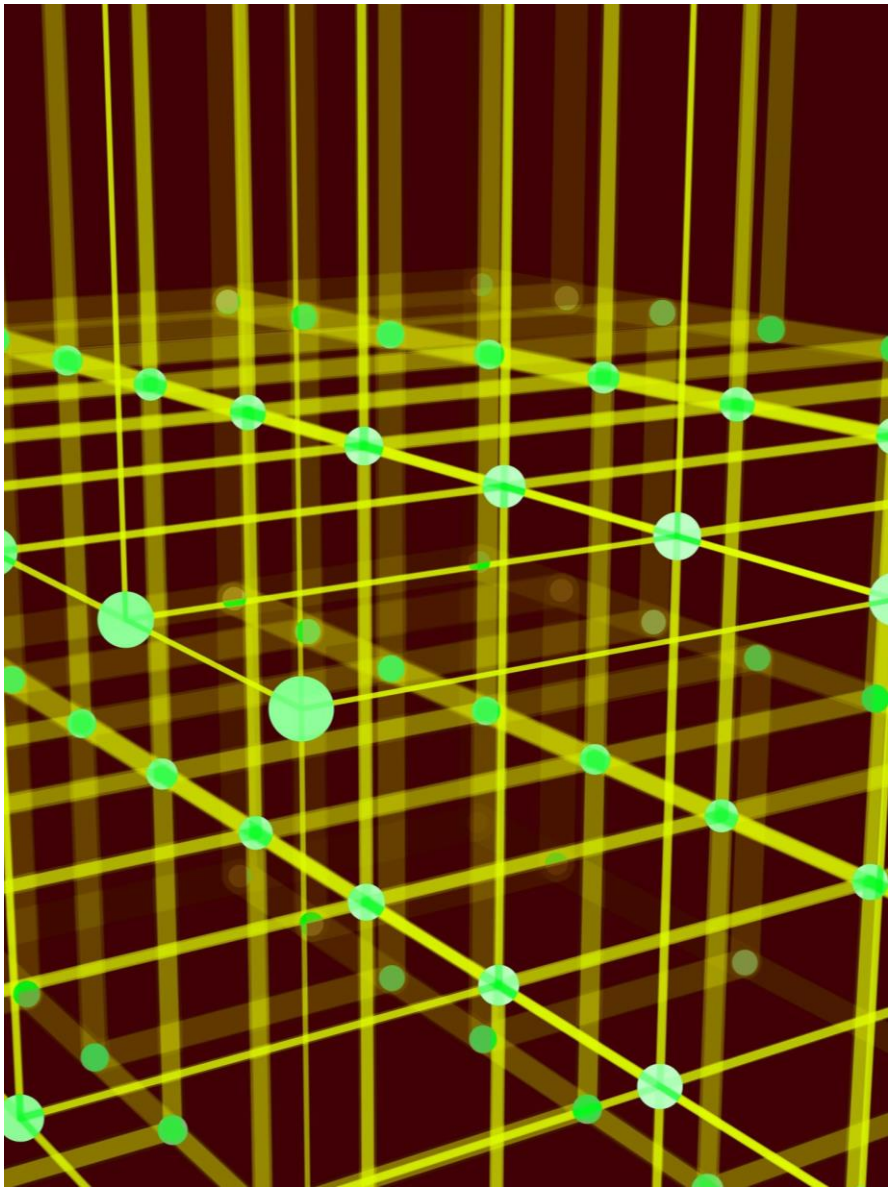
Paediatric respiratory and LTV consultant



Outline

- Explore assessment and management of respiratory disease in SMA
  - Prevention
  - *Secretion management*
  - Acute exacerbations
  - Ventilatory anticipation + management

Guidelines for management in SMA

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NMD

www.elsevier.com/locate/nmd

Diagnosis and management of spinal muscular atrophy: Part 1: Recommendations for diagnosis, rehabilitation, orthopedic and nutritional care

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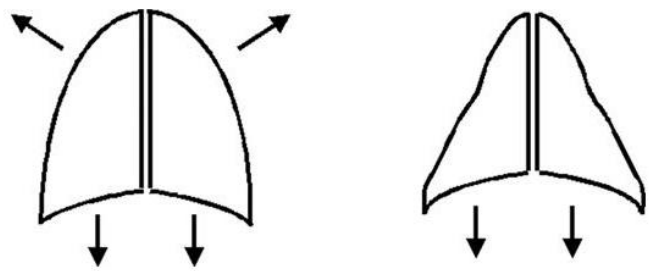
Post diagnosis initial assessment

- Early referral: non-sitters (type-1), sitters (type -2)
- Physical examination – very weak intercostals – relative stronger diaphragm
- Bell-shaped chest and pectus deformity – lung underdevelopment
- Bulbar weakness - dysphagia
- Children with SMA-1 may have disproportionate weakness of expiratory muscles, and coughing and secretion clearance needs to be evaluated

FIGURE 1



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Chest wall changes. Left, Normal lungs; right, lungs of a patient with SMA.

Spirometry

- Decreased FVC predicts frequency of chest infections
- FVC < 60% - associated with SDB
- FVC < 40% - associated with NH
- Peak cough: 270mls (>12y)
- MIP + SNIP : measures of respiratory muscle strength however convey little added benefit to FVC – more complicated tests

What about sleep?

- Sleep exaggerates instabilities in breathing pattern
- Induces hypotonia in the upper airways
- Progressive pharyngeal muscle relaxation: increases upper airway resistance (230%)
- NREM sleep: intercostal and diaphragmatic muscle contribution increases (34% and 11%)
  - Substantial increase in rib cage contribution to Vt
- REM Sleep: marked decrease in rib cage contribution ($p<0.05$) with increase (34%) of diaphragmatic muscle activity to maintain Vt

Assessment

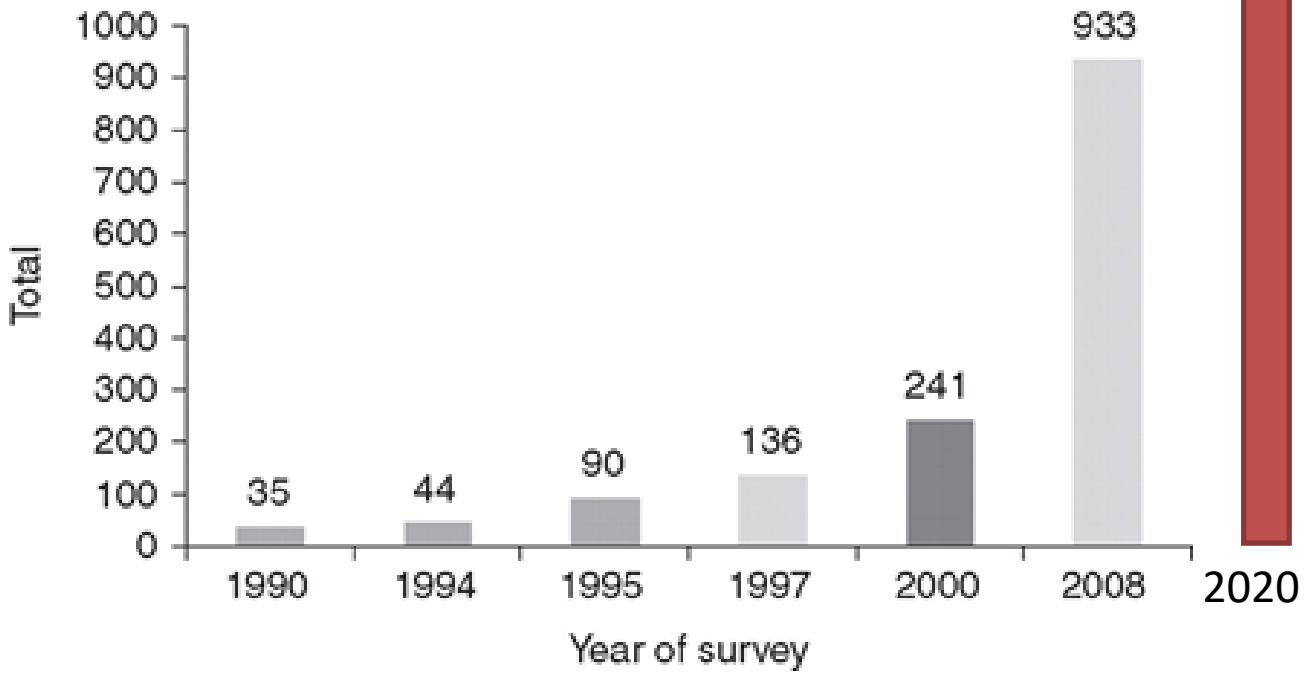
- Clinical cough effectiveness
- Overnight oximetry (minimum), PSG – ideal
  - CO2 monitoring and detection of hypoventilation
- Respiratory support
  - NIV in all symptomatic infants:- prior to respiratory failure
  - Prevent chest wall deformity
  - Aid respiratory function
  - Palliate dyspnoea
  - NO CPAP: -*ideally*
  - Careful with oxygen
  - Interface issues + compliance
  - ?tracheostomy

"Now, sleep like you would at home!"

Use of NIV

>2000

Review of current (2008) and previous surveys (published1 and personal communication) of ventilated children in the UK by year of survey (x axis) and total identified (y axis).



2020

C Wallis et al. Arch Dis Child 2011;96:998-1002

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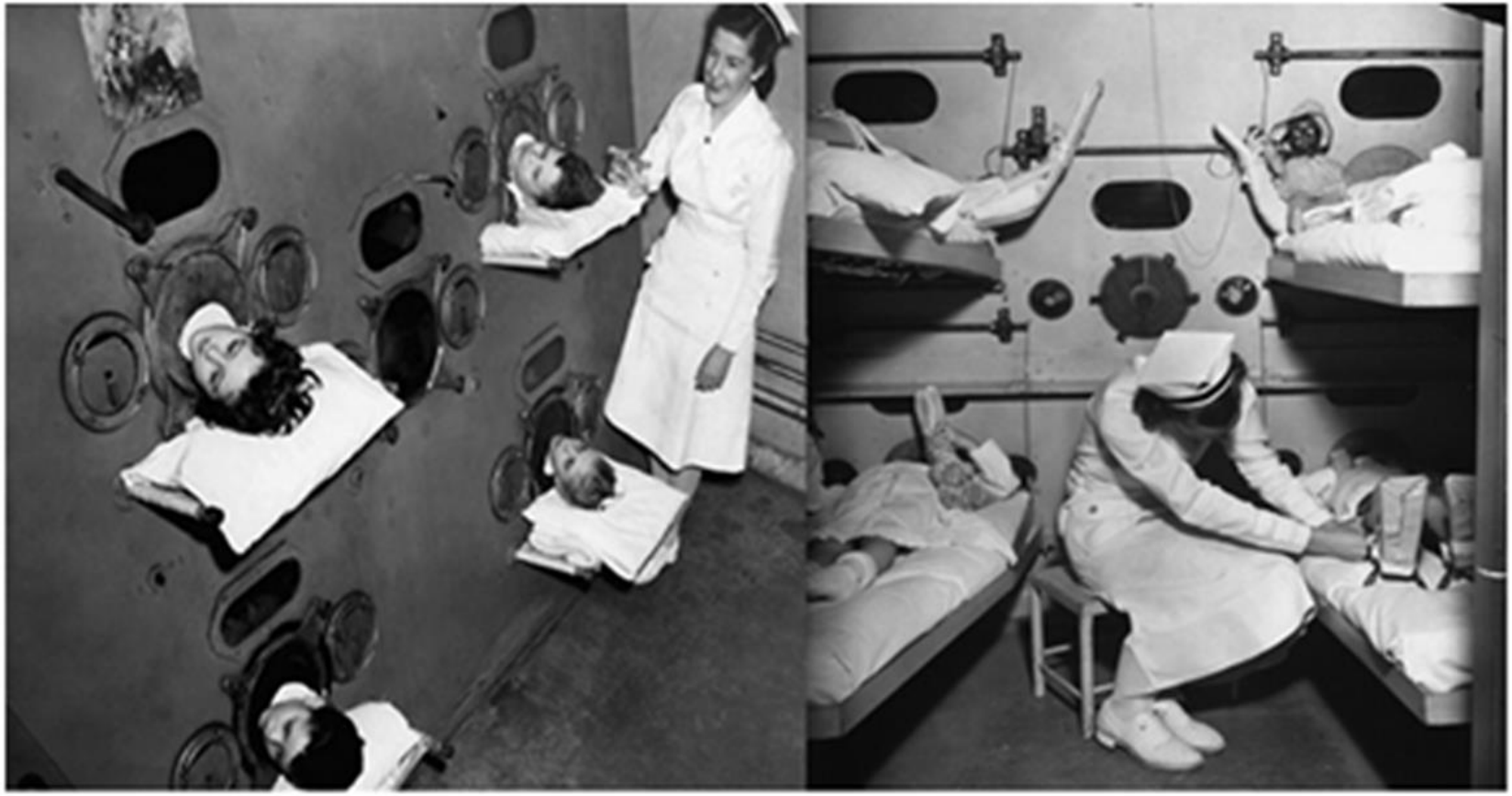
ADC

Multi-person negative-pressure ventilator at Boston Children's Hospital, 1950s.



A brief history of time

Robert M Kacmarek Respir Care 2011;56:1170-1180

(c) 2012 by Daedalus Enterprises, Inc.

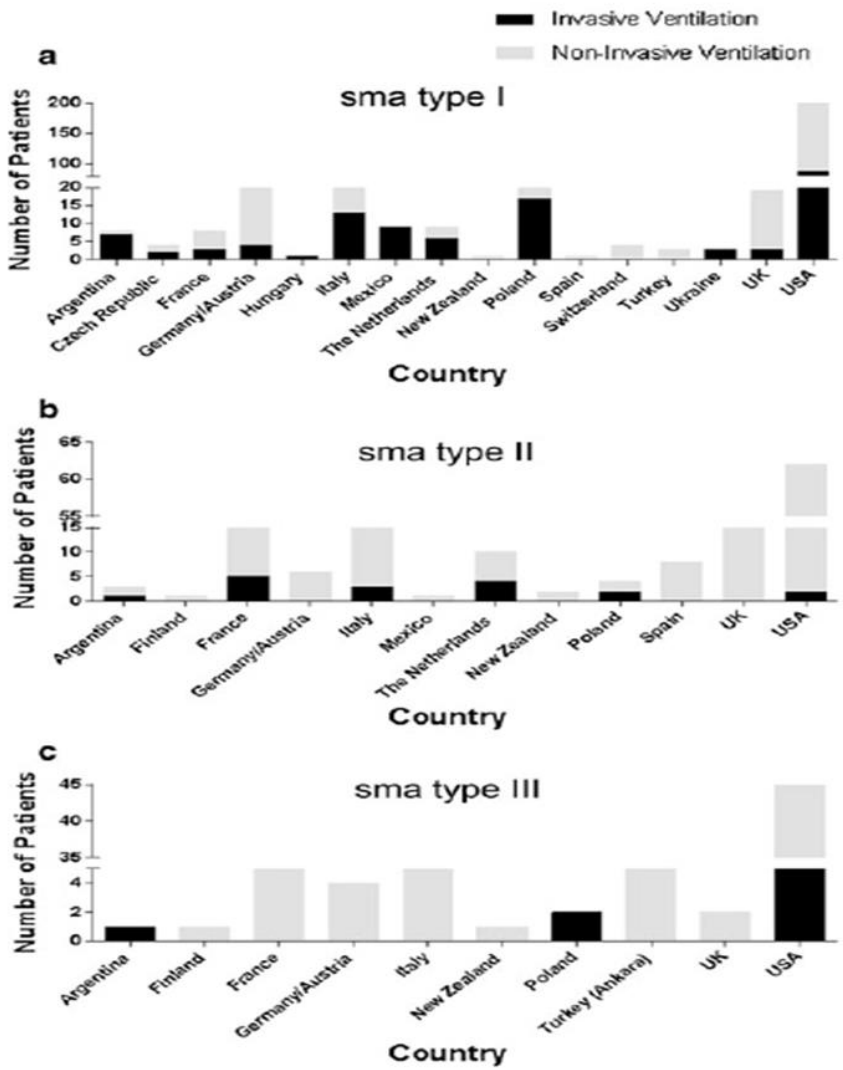
Assisted ventilation

- Ethical dilemmas – separate debate
  - Simply put: must benefit the child (guidelines)
  - Supplement in archives: “Making decisions to limit treatment in life-limiting and life-threatening conditions in children: a framework for practise” – May 2015 vol. 100 supp. 2
- Treat symptoms of NH
- Treat daytime symptoms:
  - Mechanism poorly understood
  - Muscle loading / improved hypercapnic response
- Reduce admissions secondary to infection
- Improved quality of life
- *Prolong life*

NIV interface

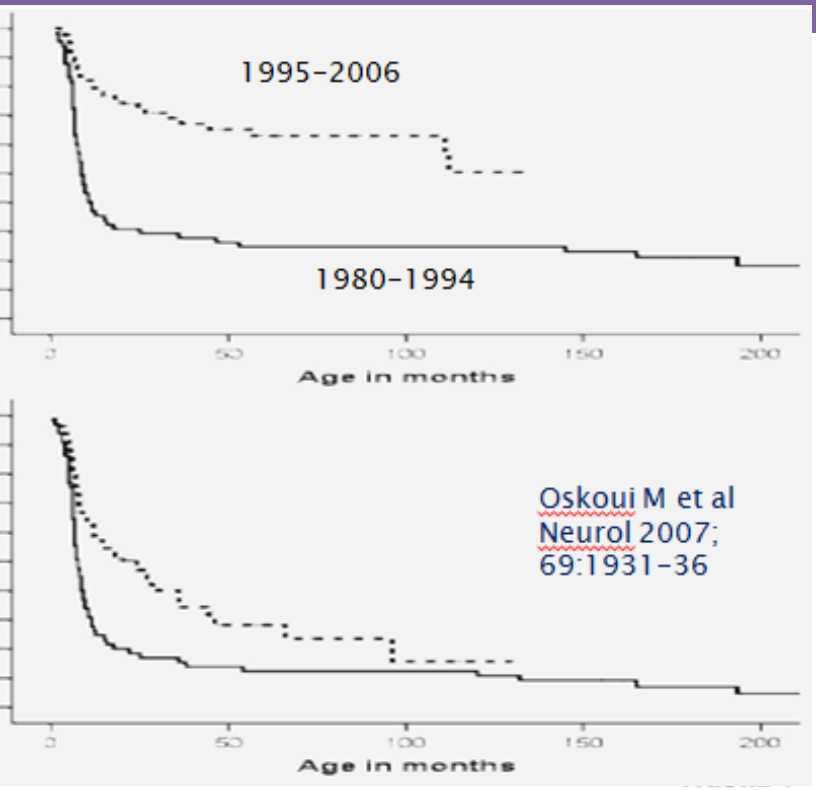
- Improvements of the years
- Mask from 4kg weight
- Currently use Breas VIVO3
  - Multiple vent options
  - Requirement to perform Bi-level support
- Low I-time
- High RR
- Low PEEP
- Clearance of CO2
- Reduce atelectasis , reduce infections, prevent hypoventialtion

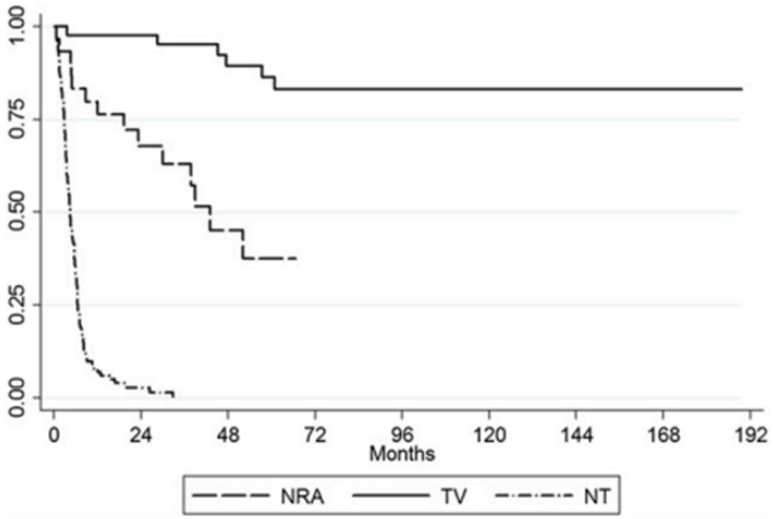
Comparison across the globe



J Neurol (2014) 261:152–163

Survival in SMA type I





Kaplan-Meier estimates of survival in the 3 groups. The cumulative probability of survival was greater for continuous NRA and TV groups, with a lower survival probability at ages 24 and 48 months in the NRA group.

Initiation of NIV in non-sitters

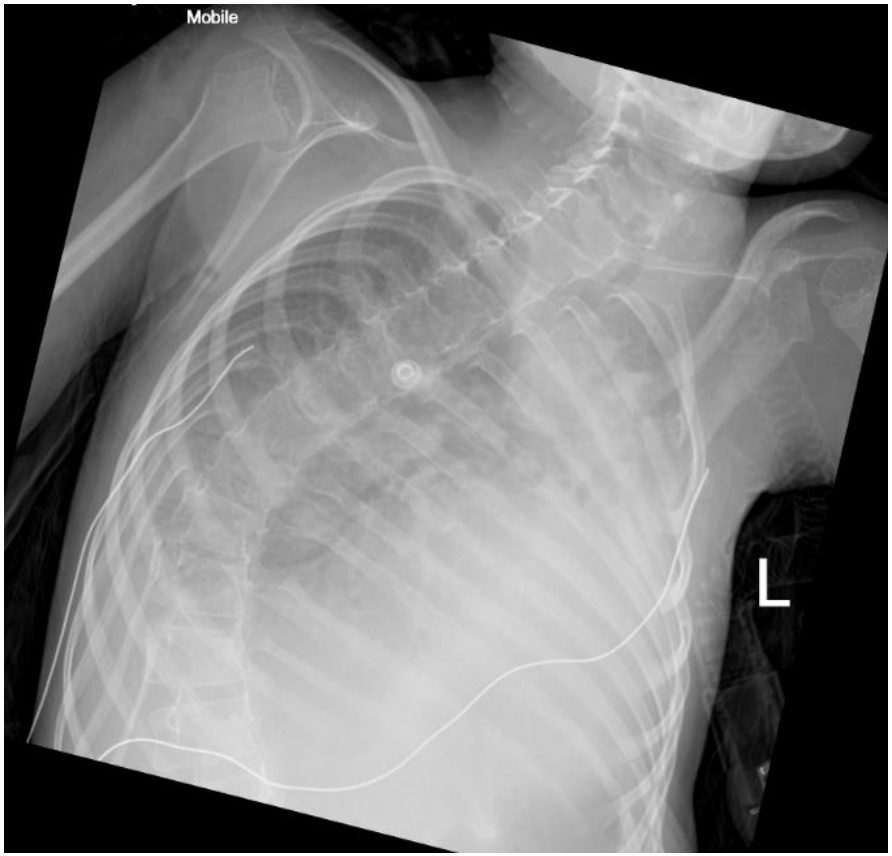
Main criteria	Supportive criteria
1. Infective exacerbation (acute) 2. Recurrent chest infections 3. Baseline increased work of breathing/ dyspnoea 4. Documented respiratory failure (either chronic (as shown on sleep studies) or acute)	1. Poor weight gain in spite of optimised feeding 2. Chest deformity

≥ 1 main criteria +/- supportive criteria

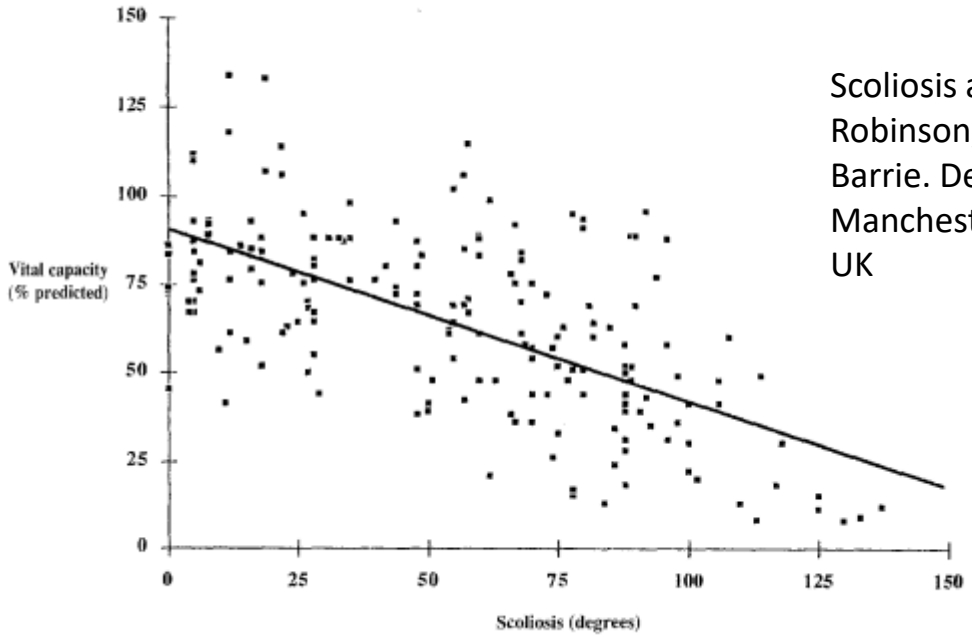
Note: supportive criteria alone are not sufficient indications for initiation of NIV

Sitters (SMA-2)

- Similar assessment process:- overnight / swallow / secretions / infection frequency + scoliosis
- Common feature – general reflects severity of associated weakness
- DMD – 70-90%, SMA I/II – 100%
- Lateral curvature:
  - Displacing thoracic cage
  - Displacing diaphragm
  - Limiting Vital capacity
  - Asymmetric inspiration
  - Lung hypoplasia (dependant on onset)
  - CO2 retention



Impact of scoliosis



Scoliosis and lung function in spinal muscular atrophy. D. Robinson, C. S. B. Galasko, C. Delaney, J. B. Williamson, and J. L. Barrie. Department of Orthopaedic Surgery, University of Manchester, Royal Manchester Children's Hospital, Manchester, UK

Fig. 3. Correlation between vital capacity (measured as a percentage of the predicted value) and scoliosis (measured in degrees using the Cobb method). There is a 4.7% reduction in vital capacity for every 10° increase in curve

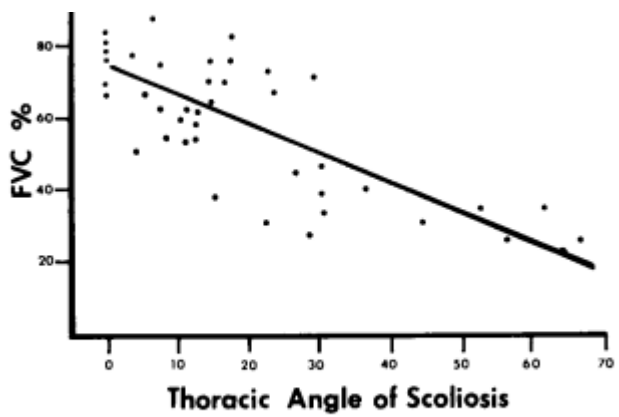


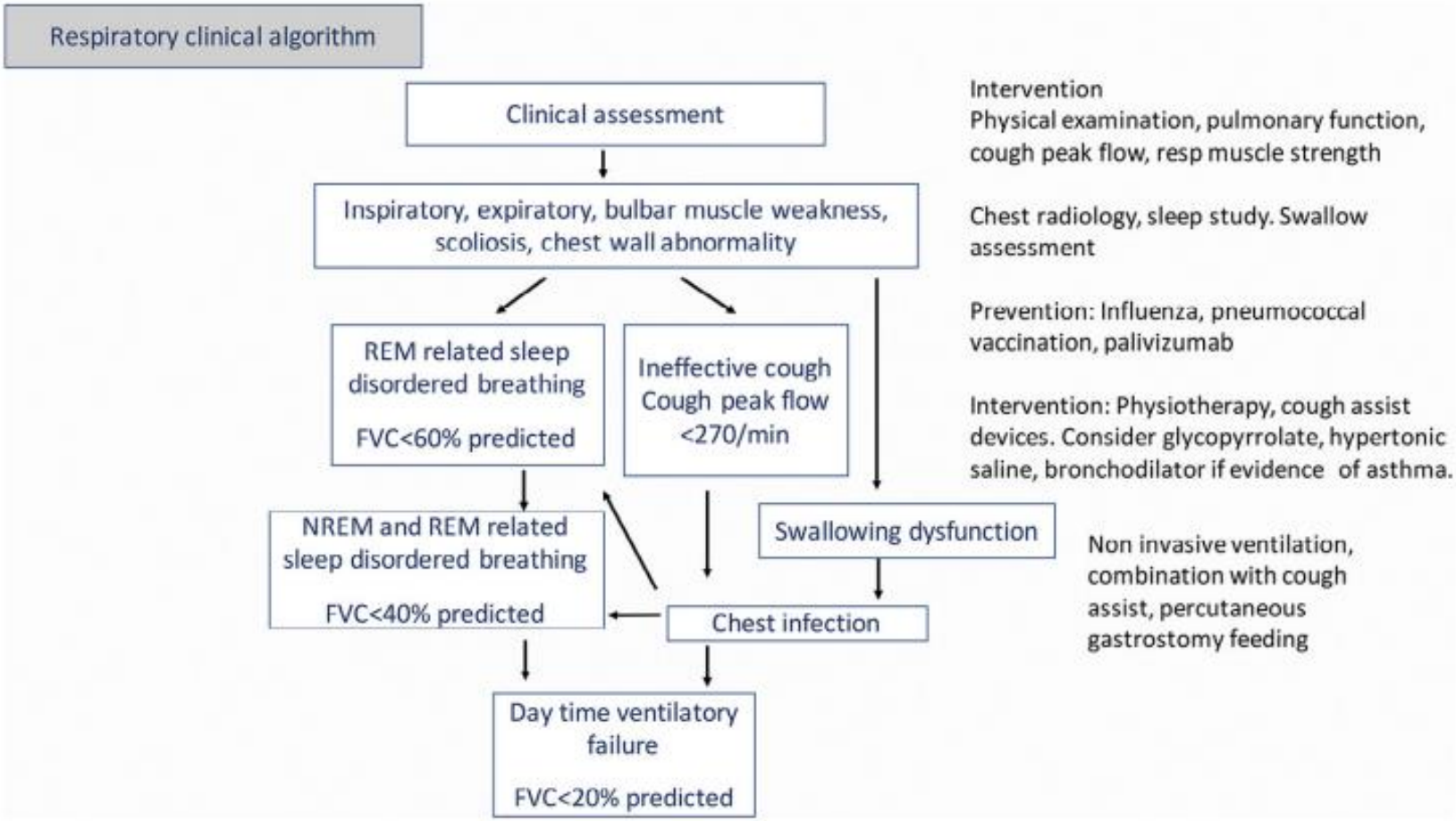
FIG. 3. Percent of predicted normal forced vital capacity (FVC) versus thoracic angle of scoliosis. The correlation coefficient r is −0.764.

- Age and the angle of thoracic scoliosis (but not lumbar scoliosis) predicted vital capacity, with each 10^0 of thoracic scoliosis being equivalent to loss of 4% of predicted vital capacity

Kurz LT, Mubarak SJ, Schultz P, et al. Correlation of scoliosis and pulmonary function in Duchenne muscular dystrophy. J Pediatr Orthopedics 1983;3:347e53.

Medications

- Antibiotics (prophylactic / therapeutic)
- ?bronchodilators
- Mucolytics + Dornase:- no evidence
- Care with hyoscine / glycopyrrolate
- Botox: - no consensus , safer to avoid
- Palivizumab: - YES
- GORD: - look for and treat


Respiratory clinical algorithm
Clinical assessment
Inspiratory, expiratory, bulbar muscle weakness, scoliosis, chest wall abnormality
REM related sleep disordered breathing
FVC<60% predicted
Ineffective cough
Cough peak flow <270/min
NREM and REM related sleep disordered breathing
FVC<40% predicted
Swallowing dysfunction
Chest infection
Day time ventilatory failure
FVC<20% predicted
Intervention
Physical examination, pulmonary function, cough peak flow, resp muscle strength
Chest radiology, sleep study. Swallow assessment
Prevention: Influenza, pneumococcal vaccination, palivizumab
Intervention: Physiotherapy, cough assist devices. Consider glycopyrrolate, hypertonic saline, bronchodilator if evidence of asthma.
Non invasive ventilation, combination with cough assist, percutaneous gastrostomy feeding

Flow chart

Long-term plans

- Set goals of care
- Respiratory care protocols
- End of life plans
- ACPs
- Intubations
- Extubation criteria

Tracheostomy
+ LTV

Tracheostomy + LTV

- We now live in an era of LTV
  - Cannot go back
  - Well publicised
    - 18 such cases considered by judges 2019
  - Easy to research
  - Parents exposed to other children on LTV
  - Communication era boomed…
  - Doctors no longer felt to be “gatekeepers of knowledge”
  - Have to work with this not against it
  - Childs interest always first: MDTs / joint decisions / mediation

PUGH

'The blasted badgers have moved the goalposts again!'

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