BTS Guideline for Respiratory Management of Children with Neuromuscular Weakness
Appendix S1
Evidence tables

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Abi Daoud M, J Dooley, K Gordon	Depression in parents of children with Duchenne Muscular Dystrophy	qualitative	2004 Pediatric Neurology 31,1:16-19	2	42	42/60 (70%) parents from 26/34 males with Duchenne MD - median age13.5 yrs (11-19) 23/27(85%) nonambulant	Case control design with frequency matched control parents in 1:4 ratio using established questionnaires Depression scale (short form CIDI-SF)	Results from Control parents from National Population Health Surveys data set	N/A	Depression scale (short form CIDI-SF)	Parents were more likely to suffer a major depressive episode than controls ($p<0.0001$) Depression score of ≥ 3 (50% probability or higher) in 31% study parents vs 4% controls, OR :10.3, (95%CI 3.5,31.6)	Summer studentship Dalhousie medical school and IWK Health centre	Almost 1/3 parents were at risk of a major depressive episode compared with 4% control parents. Parents of older males and single parents were at greatest risk.

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Abresch R, G Carter, M Jensen, D Kilmer	Assessment of pain and health-related quality of life in slowly progressive neuromuscular disease	qualitative	2002 Am J of Hospice and Palliative care	3	811	Adults (av age : 53yrs SD=15;range 18-88) post polio syndrome (n=355) SMAII (n=68), SMAIII (n=29), CMT (n=232), FSHD (n=64), MMD (n=33), LGMD (n=31) Socio-demographic data	Questionnaire : SF-36 survey (validated quality of life measure) Global ratings of satisfaction with general aspects of health, social status, activities	Comparison with data from general US population	N/A	SF-36 survey measuring 8 quality of life constructs	76% participants reported some pain, 54% reported moderate to very severe except the SMA group which was comparable to general population. Significant correlation between pain and each of the other SF-36 scales and moderate (.70>r>.30) correlation with global satisfaction of general health	Research and training Center Grant – National Institute on Disability and rehabilitation, Washington, DC	Frequency and severity of pain reported in NMD (except SMA) significantly greater than US population Significant correlation between pain and lower levels of general health, vitality and social function

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Ahlstrom G, B. Lindvall , S. Wennebe rg, J.G Gunnarss on	A comprehens ive rehabilitatio n programme tailored to the needs of adults with muscular dystrophy	Qualitative	2006 Clinical Rehabilitati on	2	37 adults in study group 39 in control group	2 counties across Sweden. Study group - 37 adults : 21 women and 16 men (mean age 50 years) control group : 39 adults - 25 women, 14 men, mean age 46 years	Comprehensi ve Rehab programme to study group consisting of 4 sessions, 10 days over 18 months	Between a study group receiving the intervention and a control group	18 months	Activities of daily living staircase Mental adjustment to Cancer scale Sickness Impact profile Psychosoci al Wellbeing questionnai re	No significant difference in outcome measures Increased dependence on others in ADL more pronounced in control group Increased trend to maladaptive coping patterns in control group	Swedish Associati on of the Neurolog ically Disabled	Information and support appears to be beneficial.

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Aloysius A., Born P., Kinali M., Davis T., Pane M., Mercuri E.	Swallowing difficulties in Duchenne muscular dystrophy: indications for feeding assessment and outcome of videofluroscopic studies.	Cross sectional	2008 Eur J Paediatric Neurol	3	30 patients aged 6-31 years	All with DMD	History of feeding issues. Evaluation of feeding assessments and videofluroscopy swallow studies (VFSS)	Between history, feeding assessments and radiological studies	Not applicable	Reported swallowing difficulties are not always associated with difficulties on VFSS	Not applicable	Not stated	Prolonged chewing and effortful bolus transport increased with age. Post swallow pharyngeal residue was common and increased with age. Feeding problems in this group of patients may go unrecognised.

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Bach, J. R., D. I. Campagnolo and S. Hoeman	Life satisfaction of individuals with Duchenne muscular dystrophy using long-term mechanical ventilatory support	qualitative	1991 Am J of Phys Med and Rehab	3	82 patients 273 controls	82 ventilator assisted patients with Duchenne MD 27.36yrs(14-44). Ventilator assisted 6.09yrs (1-21)39 NIV, 33 tracheostomy 273 healthcare professionals (controls) – mean age 31 yrs (21-59)	Questionnaires	Responses from Patients with controls And figures from general population (Campbell)	N/A	Life domain satisfaction measures and Semantic Differential Scale of General Affect (Campbell)	10/80 patients expressed dissatisfaction with life in general compared 9% controls, 7% general population. Controls estimation of patients life satisfaction 2.5+/-1.35 ($p<0.0005$)		Healthcare professionals significantly Underestimated patients scores on life satisfaction and general affect, but overestimated negative responses with ventilator use.

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Bach J.R,	Ventilator use by muscular dystrophy association patients	Qualitative	1992 Arch Phys Med Rehab	3	273	clinic/ co directors of institutes where 80 DMD ventilator users were registered across the US	questionnaire exploring reasons around offer of ventilator management brief question to users on patient satisfaction (n=80)	N/A	N/A	10 questions to directors 11th question to ventilator users with DMD	61%directors routinely ordered ventilator equipment usually when the patient was in acute respiratory failure. 143 clinic directors didnt discuss ventilator use major reason given : poor quality of life,(n=92)	no funding reported	Ventilator offer appeared to be based upon directors opinions rather than informed choice which is current accepted practice

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Bach JR	A comparison of long-term ventilatory support alternatives from the perspective of the patient and care giver	Qualitative	1993	3	Questionnaires sent to 974 long term ventilator users – not clear how they were identified. 621 responded (64%). 168 were over 18 and had used both trachy vent and NIV for at least 1 month	All used long term ventilation. 111 used trachy, and had previously used NIV for 13.7 (+/- 11.5 years). 59 used NIV having previously used trachy for for 1.6 (+/- 4.8 years. All patients used ventilation every night and at least part of the day.	Patient preference for mode of ventilation	Questionnaire responses	Single questionnaire	Questionnaire responses	Of the 111 pts initially using NIV, but now with using trachy, 55 preferred NIV and 45 preferred tracheostomy and 14 had no preference. Of the 59 patients previously managed with trachy, now using NIV, all preferred NIV Patients with trachy required routine tracheal suctioning a mean of 7.6 ±8.3 times per day. 35% percent of the patients using tracheostomy IPPV at the time of the study expressed the desire to return to noninvasive aids, whereas none of the respondents using noninvasive aids wished to switch back to tracheostomy IPPV	Not stated	Suggests patients prefer NIV. Interesting that of the 111 using trachy, 42 said that they preferred trachy. None using NIV who had previously had a trachy wanted to switch back to having one. It may be perceived that trachy represents disease progression and that may be part of the reason for avoiding it.

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Bach, J. R. and L. R. Saporito	Criteria for extubation and tracheosto my tube removal for patients with ventilatory failure	Qualitative	1996 Chest	3	49	49 consecutive patients with neuromuscular respiratory insufficiency (diagnosis include spinal cord injury, neuromuscular disease, guillain Barre, obesity, partial lung resection)	Ventilator weaning , extubation or tracheostomy tube removal facilitated by NIV and assisted coughing as needed	Comparisons of parameters such as peak cough flow, that are predictive of successful extubation or tracheostomy tube removal	Up to 70 months	Peak cough flow Vital capacity Duration of ventilator use (NIV) post decannulati on/ extubation Ventilator free breathing time SaO2 Success/fail ure in extubation/ decannulati on	Peak cough flow is an independent predictor of successful decannulation or extubation All patients with PCF> 160L/min were successfully extubated or dcannulated. No patient with PCF<160L/min were successcully decannulated or extubated.	Not stated	Adult study Not clear about the indication for translaryngeal intubation or tracheostomy in the first place –whether for acute respiratory failure Extubation and decannulation criteria clearly set down This paper reports on weaning from invasive ventilation to noninvasive ventilation (in a **non acute** setting) A weaning regime emphasizing the use of non-invasive inspiratory and expiratory muscle aids can help to remove the need for tracheostomy

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Bach JR. Ishikawa Y. Kim H.	Prevention of pulmonary morbidity for patients with Duchene muscular dystrophy	Qualitative	1997 Chest		46	Patients with DMD age range 11-26 years old. Were initially classified as 22 Non protocol patients were referred prior to 1983 and had tracheostomies. 10 protocol users who require NIV. 14 protocol patients who had recurrent hospitalisations but no NIV	Patients with a PCF< 270 L/min were taught a protocol management consisting of maximum insufflation capacity and manual assisted coughing along with mechanical insufflation/ exsufflation for when their SpO2<95% on room air	Between protocol and non protocol amount of hospitalisations. Post protocol hospitalisations and avoided hospitalisations.	Study was retrospective.	Hospitalisations of the patient group	Patient groups dependent on non protocol and protocol Protocol: n=24 Non protocol: n=17 Hospitalisations/patient Protocol: 0.5±1.0 Non protocol: 2.41±1.84 Hospitalisations/patient/ year Protocol: 0.2±0.5 Non protocol: 2.25±4.75 Hospitalisation avoided/patient/ Protocol 1.8±1.7 Hospitalisation avoided/patient/year Protocol 0.8±1.0 Hospitalise days/patient Protocol: 3.6±8.7 Non protocol: 35.4±66.3 Hospitalise days/patient/year Protocol: 1.8±5.2 Non protocol: 21.4±37.8 p<0.005 between non protocol and protocol results	Not Reported	Although the control group had tracheostomy ventilation and cannot truly be compared to the group using NIV the outcome for hospitalisation seems to be reduced with the addition of a protocol.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Bach JR, Niranjan V, Weaver B	Spinal muscular atrophy type 1*: a noninvasive respiratory management approach	Qualitative. Retrospective cohort study	2000 Chest	3	11	Eleven children with SMA I studied during episodes of respiratory failure.	Pre-extubation preparation including optimisation of oxygenation and mechanically assisted secretion clearance, followed by post-extubation NIV. "Protocol" management: NIV with no supplemental oxygen therapy. Mechanical insufflation-exsufflation and assisted coughing when SpO2 < 95% either via endotracheal tube or via non invasive support.	Protocol management vs. Conventional management for respiratory tract infections	Seven of 11 were followed for a mean of 35 months. Max follow-up 66 months.	Successful extubation to NIV. Length of survival and successful extubations - described as extubated and not needing to be intubated on same admission.	The 11 children had 28 episodes of respiratory compromise and 48 intubations. Using the protocol resulted in 23/28 succesful extubations compared to 2/20 without the protocol. 2 children survived for 37 and 66 months without intubation, but required 24 hour nasal ventilation since 5 and 7 months. 1 child had a tracheostomy for persistent left lung collapse. 6 children managed at home for 15 to 59 months. 9 children extubated successfully via protocol conventionally managed 2 out of 20 successful extubations. I child lost to follow up.	Not stated	The study is one of a number from this group. Their approach appears to be very successful in aiding extubation in a severely compromised group. Although intercurrent chest colds may necessitate periods of hospitalisation and intubation, tracheostomy can be avoided throughout early childhood for some children with SMA type 1 with a protocol approach.

Authors	Title	Study type[1]	Year Journal	Evidence Level[2]	Numbers	Characteristics	Intervention	Comparison	Follow -up	Outcomes	Effect size	Funding	Comments
Bach JR, Bianchi C	Prevention of pectus excavatum for children with spinal muscular atrophy type 1	Qualitative research	2003 Am J Phys Med Rehabil	3	3	Infants with spinal muscular atrophy type 1	Effect of high-span NIV (peak pressure at least 10cm above end expiratory pressure)	None	3- 8 years	Chest shape	Case report of 3 children treated with hi-span NIV. Historical comparison made with a trachy ventilated child who has stiff lungs and cannot tolerate large tidal volumes	Not stated	Hi-span ventilation (NIV or trachy) may prevent pectus excavatum and may promote maintenance of larger passive lung volumes

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow -up	Outcomes	Effect size	Funding	Comments
Bach JR, Vega J, Majors J, Friedman A	Spinal Muscular Atrophy type 1 Quality of Life	Qualitative	2003 Am J Phys Med Rehab	3	67 health professionals 100 family responses	Care-givers of 53 surviving children with SMA1 and 2 deceased	questionnaire with a Likert scale	comparison with responses from parents of 30 healthy children	N/A	Quality of life estimates using 10 polar adjective pairs	significant difference in perception of quality of life for SMA1 children care givers (n+104) at 7.8+/-0.2vs clinicians (n=67) 2.9+/- 0.2 (p,0.0001)	Not reported	widespread perception that SMA1 children have a poor quality of life but this perception is not shared by their care providers

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow -up	Outcomes	Effect size	Funding	Comments
Bach JR, Saltstein K, Sinquee D, Weaver B, Komaroff E	Long-term survival in Werdnig–Hoffmann disease	Qualitative	2007 Am J Phys Med Rehabil	3	92	All had SMA 1 defined by inability to sit or roll, plus at least one episode of resp failure requiring continuous resp support before 18 months, plus inability to receive any nutrition orally by 18 months. .	None – observational. Retrospective notes review between 1996 and 2006	Modes of respiratory support and use of tracheostomy	Mean of 5 years	Survival, hospitalisation rates, episodes of respiratory failure	Of the 92 infants with SMA1, 18 opted for no intervention and all died before 15 months (mean 9.6 months). 27 had tracheostomy, 47 were managed with NIV – 32 only during sleep, 16 for more than 16 hours/day and 9 continuously. Of these 47, 32 could communicate verbally compared to 6 of the 27 children with tracheostomy. Death rates in the trach and NIV groups were similar (5/27 and 8/47). Hospitalisation was more frequent in NIV group in first 3 years. Overall admissions after 5 years were unusual. Care providers rate QOL as good, although there are no details about what these children could actually do. 15/63(23.8%) had severe bradycardias. 13/25 males had cryptorchidism. 18/42 recurrent candidiasis.96 feeding tubes<24 mths age. 26 needed fundoplication. 50% early pubarche.3 acute pancreatitis	Not stated	Good longitudinal data. Useful to note that of those using NIV , 32/47 used it at night only. Of those having trach 25/27 lost ability to breathe without ventilator support immediately. Prolonged survival SMA 1 results in high incidence of comorbid medical conditions requiring addressing.

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Bach, JR. Bianchi C. Vidigal-Lopes M. Turi S. Felisari G.	Lung inflation by glossopharyngeal breathing and 'air stacking' in Duchene muscular dystrophy	Qualitative research	2007 Am. J. Phys. Med. Rehbil.	3	78	Male patients all had DMD and declining VC. Mean age= 20 ± 3.1 years.	Glossopharyngeal breathing and maximum insufflation capacity	Effect of 1) Glossopharyngeal breathing and maximum insufflation capacity on measurements of VC, MIC and GPB 2) effect of glossopharyngeal breathing on ventilator free time	7-169 months	Pre and post VC measurements via spirometer.	95% of patients were able to achieve maximum insufflation capacity VC increased from 987 ± 631 to 1501 ± 618mls, unassisted PCF from 145 ± 112 to 250 ± 84L/min. 27 % were able to achieve glossopharyngeal breathing. Glossopharyngeal breathing increased VC from 244 ± 151 to 824 ± 584mls (P<0.001) and unassisted PCF from 164 ± 76 to 289 +- 91L/min (p<0.001). Patients who could glossopharyngeal breathe required 1.9 fewer ventilator assisted breaths per minute	Not stated	The ability to increase lung volumes with a maximum insufflation capacity is retained better than trying to increase lung volume with glossopharyngeal breathing over a period of time. Fifteen patients could glossopharyngeal breathe sufficiently to have a greater ventilator free time.

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Bach JR. Mahajan K. Lipa, B. Saporito L. Goncalves M. Komaroff E.	Lung insufflation capacity in neuromuscular disease	Qualitative research	2008 Am. J. Phys. Med. Rehbil.	3	282	Patient diagnosis and number of patients(n=), age range: DMD (n=53) 14-44 Myotonic (n= 6) 36-53 other myopathies (n=55) 11-85 SMA (n=31) 7-56 ALS (n=76) 27-82 Post polio (n= 25) 16-58 Miss (n=36) 7-85	Maximum insufflation capacity and passive lung capacity via a lung recruitment bag. When VC is < 60% predicted	Changes in VC and PCF with maximum insufflation capacity and lung insufflation capacity	one off visit	PCF VC	Baseline VC: 1131 ±744ml Maximum insufflation capacity: 1712 ± 926ml Lung insufflation capacity: 2069 ± 867 ml, Baseline unassisted PCF: 2.5 ± 2.0L/sec Assisted PCF: 4.3 ± 2.2 Changes from baseline were significantly different ($p<0.01$)	Not stated	Benefits can be greatest for most advanced patients with lowest VC's. Regular lung insufflation, either by air stacking or by passive lung insufflation is indicated for all patients with NMD and diminishing VC. Passive insufflation is used when patient obtains a deeper volume in this manner by air stacking. Often beneficial to prescribe both methods

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Bach J.R, M. Gonzalez , A. Sharma, K.Swan and A. Patel	Open gastrostomy for noninvasive ventilation users with neuromuscular disease	Qualitative	2010 Am J of Phys Med and Rehab	3	62 adults	44 with amyotrophic lateral sclerosis, 10 DMD, 8 other conditions all NIV users with FVC<40% predicted.	open gastrostomy for ventilator users, without tracheostomy, intubation or general anaesthesia		N/A	reported results including complications	all patients subsequently gained weight. No procedural complications. mean post gastrostomy survival 38.8+/- 6.2 months.	not reported	An adult study - not currently applicable to the paediatric population although procedure described in NIPPV users

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Bagnall A, Mohammad A. Al-Muhaizea, Manzur A	Feeding and speech difficulties in typical congenital Nemaline Myopathy	Qualitative	2006 Advances in Speech-Language Pathology	3	19	Children with Nemaline myopathy 6 males and 13 females Aged 1y 6mo to 28 y (n=15 under 18yrs) 18 ambulant 9/19 required NIV	Retrospective notes review Investigating feeing and speech difficulties	N/A	N/A	Descriptive Growth, respiratory status	14/19 had faltering growth (defined) 11/19 had recurrent respiratory infection in 1st 3 years (61%) Reduced chest infection after gastrostomy in 4 .	Not reported	Feeding difficulties and faltering growth common, often requiring gastrostomy followed by NIV.

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Bayar, B., F. Uygur, K. Bayar, N. Bek and Y. Yakut	The short term effects of an exercise programme as an adjunct to an orthosis in neuromuscular scoliosis	Qualitative	2004 Prosthetics and orthotics international	3	15	15 patients with neuromuscular disease aged 9-16 yrs 4 Friedreich Ataxia 3 Spinocerebellar degeneration 3 congenittal muscular dystrophy 3 congenital myopathy 3 CMT	Exercise programme including postural training, muscle strengthening and stretching exercise Wearing orthosis	Changes in muscle strength before and after exercise Effect of orthosis on FVC	Intervention lasted 4 wks	Lung function Muscle strength and balance Cobb angle	FVC decreased by 18% upon wearing orthosis which deceased by 9.28% after therapy Increase in muscle strength post treatment was statistically sig ($p<0.05$)	Not stated	Small number Short follow up period

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Birnkrant DJ. Pope JF, Lewarski J, Stegmaier J, Besunder JB.	Persistent pulmonary consolidation treated with intrapulmonary percussive ventilation: a preliminary report.	Qualitative	Pediatr Pulmonol 1996	3	4	3 paediatric and 1 adult. 3 of the 4 have neuromuscular disease and one had segmental atelectasis due to aspiration	Treatment with IPV	Clinical and radiographic improvement.	One off hospital admission	3 out of 4 patients showed both clinical and radiographic improvement within 48 hours. Remaining patient experienced third degree AV block, hypoxemia and bradycardia during 2 sessions- safely restarted and slowly improved	A report of 4 patients who were not improving with conventional treatment. Patients CXR improved following IPV. One patient deteriorated due to movement of a large amount of secretions	Not stated	IPV appears to be safe and effective therapy for selected patients however further clinical evaluation required.

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Bimkrant D J	New challenges in the management of Prolonged Survivors of pediatric neuromuscular Diseases : A Pulmonologist's perspective	qualitative	2006 Pediatric Pulmonol	3	19	DMD patients 18/19 assisted ventilation, 16/18 NIV, 12 24hrs /day.	Descriptive study of non-cardiopulomonary complications	N/A	N/A	Description of complication and intervention	malnutrition/dysphagia (15),nephrolithiasis (6) Diabetes (2), DVT (2), gallstones (1), Inflammatory bowel disease (1)	Not reported	Discussion on effects of prolonged survival on medical decisions linked to prognosis and burden of care

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Bostrom K, Ahlstrom G	Quality of life in patients with muscular dystrophy and their next of kin	Qualitative	2005 Int J of Rehab research	3	46 patients 36 next of kin	46 patients with muscular dystrophy – 3 groupsproximal, myotonic and myopathia distalis tarda hereditara	Subjective estimation of quality of life(SQol)		N/A	Subjective estimation of quality of life (SQol)	Patients scored lower on most items than next of kin . Age of onset had an impact on QoL		Ventilator users reported lower energy but majority were satisfied with their lives.

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Bothwell J, J Dooley, K Gordon, A MacAuley, P Camfield, J MacSween	Duchenne Muscular Dystrophy – parental perceptions	qualitative	2002 Clinical Paediatrics	3	31	All Duchenne MD, aged 4.7 – 23 yrs (mean 11.4)attending neuromuscular services in Canada	Questionnaires to parents regarding importance of services, health issues and quality of life issues	N/A	N/A	Questionnaire on services offered, research and other quality of life challenges	Physiotherapy rated as most important now (77%) and future (90%).OT (61% vs 76%) Respirology (42% vs 69%) Physical health and mobility was esp important to younger boys whilst mental health issues important in older boys	Not reported	Paediatricians need an awareness of both immediate concerns and increasing requirement to address social needs as boys become older..

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Brito, MF. Moreira, GA. Pradella-Hallinan M. Tufik S.	Air stacking and chest compression increase peak cough flow in patients with Duchene muscular dystrophy	Qualitative research	2009 J Bras Pneumol	3	30 included but 2 excluded as they could not comply with instructions therefore results are from 28 patients with DMD	DMD FVC (= 29 ±12% predicted Mean age 20 +/- 4 years. All patients were receiving NIV. All patients were stable and free of RTI's	Patients underwent the following cough augmentation methods in a random order: Manual assisted cough, Maximum insufflation capacity, Combination of maximum insufflation capacity with a manual assisted cough	Baseline unassisted PCF to PCF with Manual assisted cough, Maximum insufflation capacity, Combination of maximum insufflation capacity with a manual assisted cough	One off visit	Effect of Manual assisted cough, Maximum insufflation capacity, Combination of maximum insufflation capacity with a manual assisted cough On PCF	Baseline unassisted PCF 171± 67L/min Manual assisted PCF 231± 81 L/min Maximum insufflation capacity 225±80 L/min Combination of maximum insufflation capacity and manual assisted cough 292± 86L/min There was a significant increase from baseline unassisted PCF with each cough augmentation technique ($p<0.001$) there was also a significant difference between each intervention ($p<0.05$)	Association for the incentive funding of psychopharmacology	Both manual assisted cough and maximum insufflation capacity were efficient in increasing PCF. However the combination of these two techniques had a significant additional effect.

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Brooke MH, Fenichel GM, Griggs RC, Mendell JR, Moxley R, Florence J, King WM, Pandya S, Robison J, Shierbecker J,	Duchenne muscular dystrophy: patterns of clinical progression and effects of supportive therapy	Cohort study	1989 Neurology	3	293	283 boys with DMD and 10 with Becker MD.	None	Muscle strength and rate of progression of scoliosis	Median of 3.6 years. 109 patients followed for more than 5 years.	89 of 120 patients developed a scoliosi	No beneficial effect seen from use of bracing. Numbers treated are not mentioned, The only statistical correlation was a positive correlation between severity of scolioisandnumber of hours per day spinal brace was wron.	Not stated	Descriptive study only, with limited hard data.

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Brooks D., A. King, M. Tonack, H. Simson, M. Gould, R Goldstein.	User perspectives in issues that influence the quality of daily life of ventilator assisted individuals with neuromuscular disorders	Qualitative	2004 Canadian Resp. J	3	26	Canadian ventilator assisted individuals with NM disease(mean age 44+/-14 yrs) Mean ventilator experience 18+/-13 yrs, 16 male and 10 female All living in the community	To identify issues that impact on daily life – use of semi-structured interviews	N/A	N/A	Semi-structured interview based upon 14 questions	Quality of life rated as mean 8.1/10. Described period of adjustment to the ventilator, HMV considered a key contributor to energy and ability to participate. Dependence on others was a negative contributor esp. suctioning		Useful perspective and suggestions as to how services could be improved

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Brouillette R.T., Morielli A., Leimanis A., Waters K.A., Ruciano R., Ducharme F.M.	Nocturnal pulse oximetry as an abbreviated testing modality for pediatric obstructive apnea	Cross sectional study	2000 Pediatrics	3	349 subjects	Referred for possible obstructive sleep apnoea	None	Nocturnal oximetry data compared to polysomnography data	None	Oximetry alone cannot distinguish between obstructive apnoea and hypoventilation	Not applicable	From local hospital foundations and SIDS Research Groups	This study did not consider children with neurodisability. However, it is likely that oximetry alone is equally unable to distinguish between types of respiratory events in neuromuscular disease.

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Brown, J. C., J. L. Zeller, S. M. Swank, J. Furumasu and S. L. Warath 1989	Surgical and functional results of spine fusion in spinal muscular atrophy	Qualitative research	1989 Spine	3	84	84 patients with SMA seen at a single centre from 1965-87	27 patients excluded Group I: 34 had posterior spinal fusion with Harrington rod Group II: 6 patients had posteriori fusion with Luque segmental spinal instrumentation	Complication rate Evaluations in ambulation, equipment use, and functional activities pre and post op	Group I follow up 4-19 yrs Group II follow up 3.5 yrs	Complication rate Physical and occupational therapy evaluation pre and post op	Gp I complication rate 35% average pre op curve 57 ^{o} average post op correction 42% Gp II Complication rate 16% average pre op curve 37 ^{o} average post op correction 42% No difference in function between the 2 operative gps.	Not stated	This study describes a single centre experience performing spinal surgery spanning 2 decades using techniques which are much less commonly used in present days.

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Brunherotti, M. A., C. Sobreira, A. L. Rodrigues-Junior, M. R. de Assis, J. Terra Filho and J. A. Baddini Martinez	Correlations of Egen Klassifikation and Barthel Index scores with pulmonary function parameters in Duchenne muscular dystrophy	Qualitative	2007 Heart Lung	3	29 patients	All DMD, 26 were able to complete study. Mean age 12.7 years (range 7-22)	Measurement of spirometry and blood gases. Assessment of twofunctional scales, the Egen Klassification (EK) and the Barthel Index (BI).	Each index was related to lung function parameters and to blood gas results.	Not applicable	Functional scales can predict spirometry and patients at high risk of needing ventilation.	EK related to FVC -0.751. BI related to FVC 0.679	Not stated	The functional scales correlated less well with FEV1 than with FVC.

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Cambridge W, Drennan JC	Scoliosis associated with Duchenne muscular dystrophy	Retrospective cohort study	1987 Journal of Paediatric Orthopaedics	3	105	Patients with DMD. !3 patients were braced and 14 went on to spinal surgery	None	None	Follow up for operated patient 25-125 months Not stated for those who had orhtosis	Non specific	Bracing was unsuccessful in 94%. The degree of kyphosis was predictive of loss of ambulation and life expectancy	Not stated	A descriptive paper of the progression of scoliosis in a cohort of patients from a single unit

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Chatwin, M Ross E. Hart N. Nickol, AH. Polkey M I. Simonds AK	Cough augmentation with mechanical insufflation /exsufflation in patients with neuromuscular weakness	RCT	2003 ERJ	1-	22 patients, 19 control group	Aged 10 to 56 intermediate spinal muscular atrophy (n=10), Duchene's muscular dystrophy (n=6), poliomyelitis (n=3) and other congenital muscular dystrophies (n=3). 17/22 patients were using nocturnal NIV at the time of the study Patients had required between two and six courses of antibiotics during the preceding 12 months for respiratory tract infections and all reported poor cough.	4 cough augmentation methods performed in a random order. 1) manual assisted cough 2) NIV assisted cough 3) exsufflation assisted cough 4) mechanical insufflation exsufflation assisted cough.	Changes in PCF from baseline for the 4 following cough augmentation techniques1) manual assisted cough 2) NIV assisted cough 3) exsufflation assisted cough 4) mechanical insufflation exsufflation assisted cough. Comparison between the NMW group and controls for respiratory muscle strength and PCF	One off visit	Primary outcome measure PCF. With comparisons between respiratory muscle strength FEV1, VC, SpO2 and EtCO2. VAS for feedback regarding each technique.	Unassisted cough 169 (129± 209) L/min; manual assisted cough 188 (146± 229) L/min; non-invasive ventilator-assisted cough 182 (147± 217) L/min; exsufflation-assisted cough 235 (186± 284) L/min; and mechanical insufflation exsufflation assisted cough 297 (246± 350) L/min. There was a significant improvement only with mechanical insufflation exsufflation from baseline unassisted PCF in the NMW group ($p<0.001$). Respiratory muscle strength and baseline SpO2 was significantly lower in the RMW group compared to their healthy controls ($p<0.005$).	MC was supported by Jennifer Trust for SMA. ER was supported by the CF Trust AHN by the BLF and NH by Association Francaise Contre Les Myopathies	Present data confirms that a combination of mechanical insufflation-exsufflation increases PCF in both children and adults with severe respiratory muscle weakness secondary to childhood onset neuromuscular disease, in the absence of severe bulbar dysfunction. Further studies recommended looking at the long term outcome of mechanical insufflation-exsufflation. No adverse events reported

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Chatwin, M, Nickol A H, Morrell M J., Polkey M J and Simonds A K.	Randomised trial of inpatient versus outpatient initiation of home mechanical ventilation in patients with nocturnal hypoventilation	RCT	2008 Respiratory Medicine	1-	28 14 randomised to inpatient initiation of home ventilation and 14 to outpatient	All were over 11 years of age (mean 45) with nocturnal hypercapnia (pCO_2>6.5 for >50% of night) and symptoms of hypercapnia. Wide range of diagnoses – only 4 had DMD and 4 had SMAII. Others had old polio, diaphragm palsy, ALS, scoliosis. Of these only the ALS was progressive. 10 out of the 28 used wheelchairs	Home mechanical ventilation for patients with stable restrictive lung disease	Success of outpatient versus inpatient initiation of NIV.	12 months	Primary outcomes were daytime ABG, nightime TcCo2, night time sats. Secondary was QOL, convenience, use of health care resources	There was apparently no difference in any of the primary or secondary outcomes. One of the analyses looks like it should be different: time TcCo2 above 6.5kpa was around 90% for both groups at baseline. For the OP group this time only decreased by 3%(+/-12), whereas it decreased by 29% (+/-37%) in the IP group. The very wide variance may explain the lack of statistical difference. The wide variance also suggest that some patients got worse (ie increased time of hypercapnia). Over all although there was no difference between the groups there were rather modest effects on night time TcCO2, daytime CO2 and night time sats. Also, the average use of the NIV was only around 4 hours (+/- 2.5) hours per night. Despite this both groups reported moderate improvements in some domains of the SF36 QOL.	M.C. received educational grants from the Jennifer Trust for Spinal Muscular Atrophy UK and Breas Medical, Sweden. A.N. was supported by grant from the Royal Brompton & HarefieldNHS Trust Clinical Research Committee.	Although the patient groups were not all children, the study demonstrates that outpatient initiated NIV is as successful as in-patient initiated treatment. Children will always have carers, and so there is even more reason to think that OP initiated treatment would be successful.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Chatwin M, Bush A, Simonds A K.	Outcome of goal directed non-invasive ventilation and mechanical insufflation /exsufflation in spinal muscular atrophy type I	Qualitative	2011 Arch Dis Child	3	13	Retrospective case note review of all children referred to authors tertiary centre (Brompton) with SMA1. Patients were treated with NIPPV to correct nocturnal hypoventilation, reverse observed paradoxical breathing and facilitate airway clearance during physiotherapy	Use of 'goal-directed' NIV	None – observationsal study	Retrospective review	Management options, chest shape and survival	5 of 14 patients died. Tracheostomy was discussed and declined in all cases. IPAP was titrated to treat hypoventilation with a median IPAP of 18 cm H2O. EPAP was 4cm. None of the infants treated with NIV in this study developed pectus excavatum. The paper provided photographs of 2 infants showing pectus excavatum prior to starting NIV with considerable improvement after 12-18 months Families of the 5 infants who died reported that their child's breathing appeared more comfortable with ventilatory support. Parents reported that NIPPV enabled them to have family time with their child at home, which would not have been possible in the hospital environment. NIPPV and MI-E were used for successful protocol-led extubations (n=9) but not non protocol-led successes (n=3).	National Institute for Health Research Respiratory Disease Biomedical Unit at the Royal Brompton hospital. MC has been supported by Jennifer Trust and a unrestricted educational grant from Breas Medical Sweden	Shows NIV can be used as part of a palliative approach to SMA1. Causes of death are not given for 4 of the 5 children. It is also not clear for which children a decision was made to use NIV in a palliative fashion and for which it was part of a Bach style approach to prolong survival. MI-E is helpful in this group and was provided around the age of one year old. Protocol treatments were more successful than non protocol. High-span NIV seemed to prevent chest wall deformity.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow -up	Outcomes	Effect size	Funding	Comments
Chng SY, Wong YQ, Hui JH, Wong HK, Ong HT, Goh DY	Pulmonary function and scoliosis in children with spinal muscular atrophy types II and III	Qualitative	2003 J Pediatr Child Health	3	12	Eight children with SMA II and 4 with SMA III, all of whom had scoliosis surgery	None	Rate of progress of scoliosis; lung function following surgery	Mean of 44 months post-operatively (but wide range)	Cobb angle. FVC pre and post-operatively	The rate of decline in FVC dropped from 7.7% per year pre-operatively to 3.8% per year post-operatively.	Not stated	Very little longitudinal study of pre-operative FVC.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow -up	Outcomes	Effect size	Funding	Comments
Colbert AP, Craig C	Scoliosis management in Duchenne muscular dystrophy: prospective study of modified Jewett hyperextension brace.	Case control study	1987 Arch Phys Med Rehabil	2	22	Patients with DMD and scoliosis. Seven were given Jewett braces and there were 15 controls	Bracing to control scoliosis	Rate of progression of scoliosis	2-7 years	Cobb angle	The rate of progression was slower in the braced group (8.8 vs 11.2 degrees/year) but not felt to be clinically significant	Not stated	A good study with controls but not randomised so prone to some bias. Although a difference in rate of progression was found it was felt to be clinically unimportant

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow -up	Outcomes	Effect size	Funding	Comments
Dawson S, Kristjanson L J	Mapping the journey: Family Carers' Perceptions of Issues related to End-Stage Care of Individuals with Muscular Dystrophy or Motor Neurone Disease	Qualitative	2003 J of Palliative Care	3	16	16 family carers :DuchenneMD (n=11) and Motor Neurone Disease (n=5)	Qualitative study via in-depth interview Semi-structured focusing on end of life issues	N/A	N/A	Common themes from interviews are discussed	Transition of services from children to adults was crucial, inconsistent access to and understanding of palliative care. Respite care was essential but not available. Lack of understanding within acute care facilities		Some salutary lessons for health care settings. Need for education and coordination of approach, esp. in acute settings. Need for palliative care plans and approach

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Dohna-Schwake, C., R. Ragette, H. Teschler, T. Voit and U. Mellies	Predictors of severe chest infections in pediatric neuromuscular disorders	Qualitative, retrospective case note review	2006 Neuromuscul Disord	3	46 subjects	6-20 years old Various neuromuscular disorders: 16 Duchenne MD, 14 Spinal Muscular Atrophy, 10 Congenital MD, 3 Neuropathy, 3 Myopathy	Measurement of IVC, FEV1, FVC, Peak Inspiratory Pressure, Peak Expiratory Pressure, and Peak Cough Flow	Between various lung function measures and the occurrence of severe lower respiratory infection (LRI)	Case note review	VC < 1.1 litres correlates with infections. PIP does not predict infection. PCF < 160 l/min correlates with infections. PCF correlates with IVC. PCF correlates with PEP.	Not applicable	Not stated	This paper shows correlations between absolute measures of lung function and the risk of developing severe LRI, potentially allowing identification of groups at particular risk.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Dohna-Schwake C. Ragette R. Teschler H. Voit T. Mellies U.	IPPB-assisted coughing in neuromuscular disorders	Qualitative research	2006 Pediatrics	2-	29	School children (6-20 years) with NMD with a PCF less than 160Lmin and either 1 admission for pneumonia or 2 chest infections treated wit IVAB's hospital admission in previous year. DMD=5, congenital MD= 7, lamin2 deficiency= 4, rigid spine MD=1, Ulrich MD=1, SMA type 2=8, SMA type1b= 5, Hereditary sensor motor neuropathy =2, CMD=1, nemaline myopathy=1 and 1 late onset Pompe's. 26 had scoliosis. 17 used nocturnal NIV.	To investigate whether hyperinsufflation with an intermittent positive pressure breathing (IPPB) device is effective in increasing PCF	pre a post forced inspiratory vital capacity and PCF pre and post maximum insufflation capacity with IPPB	2 days	Spirometry-FIVC, PIP, PEP, PCF, FEV1	In 28/29 patients, IPPB-assisted hyperinsufflation enhanced FIVC from 0.68 +/- 0.40 l to an maximum insufflation capacity of 1.05 +/- 0.47 l ($P < 0.001$). Unassisted PCF was 119.0 +/- 57.7 l/min, and increased with maximum insufflation capacity to 194.5 +/- 74.9 l/min ($P < 0.001$) in 27/29 patients. This effect was similar in young patients (ages 6-10 years) and older patients (aged >10 years). Augmentation of lung volumes from FIVC to maximum insufflation capacity correlated with an increase of PCF ($R = 0.42$, $P < 0.05$). Pressures used for hyperinflation ranged from 22-40mbar	Grants from: VitalAire, Heinen und Lowenstein, University of Essen, European Union and Alfried Krupp von Bohlen	Results suggest that this technique can be used as a method of maximum insufflation capacity to increase PCF and therefore increase cough strength in children with NMD

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow -up	Outcomes	Effect size	Funding	Comments
Dohna-Schwake, C, Podlewskii P, Voit T and Mellies U.	Non-invasive ventilation reduces respiratory tract infections in children with neuromuscular disorders	Qualitative	2008 Pediatric Pulmonology	3	35 24 patients using NIV for at least 12 months and 11 not using NIV	Children with neuromuscular weakness, mean age around 12 years. 24 patients were using NIV for nocturnal hypoventilation, or because of daytime hypercapnia.	NIV. Cough assist also used.	Questionairre given to carers asking about hospital admissions for respiratory tract infection (RTI) and attendance at GP.	Single question airre.	Hospital admission data and GP attendance.	Children on NIV had at least a 50% reduction in number of hospital admissions after starting NIV. In the first year of NIV GP consultations for RTI decreased from 9.2 to 3.2 per year ($P<0.005$), the number of antibiotic treatment due to RTI decreased from 4.1 to 1.9 per year ($P<0.005$) and the number of hospital admissions due to RTI decreased from 1.6 to 0.7 per year ($P<0.005$). In 12 patients using NIV for more than 5 years the incidence of RTI requiring hospital admission decreased from 0.54/year in the pre-ventilation period to 0.12/year in the NIV period ($P<0.005$).	not stated	Study shows that children using NIV have lower rates of hospital admission than those not on NIV. Not clear how those not using ventilation had been screened for hypoventilation or symptoms. No comments on the severity of their respiratory impairment. Also 15/25 of those using NIV also used assisted cough

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Durkin E.T, M.K.Schroth, M.Helin and A.F Shaaban	Early laparoscopic fundoplication and gastrostomy in infants with SMA Type 1	Qualitative	2008 J of Pediatric Surgery	3	12	Children with SMA type 1	Retrospective records review of early laparoscopic fundoplication and gastrostomy insertion shortly after diagnosis Standardized post operative management and discharge criteria	none	Information at one year post operatively	Description of post op care, complications and follow up care	Clinical evidence of reflux in 8/12 (66%). Clinical resolution of GER post procedure. Average time to full feeding 42+/- 4.9 hours (range 30-48). Inpatient stay : 78+/- 22.5 hrs (range 44-120) Mean weight for length was doubled at one year (P=o.o3) Number of respiratory related hospital admissions decreased by almost 50% but statistical significance not reached	None declared	The number of respiratory related hospitalizations decreased by almost 50% in 12 mths after surgery but statistical significance not reached in this small cohort. One patients GER increased dramatically following previous gastrostomy insertion necessitating referral for fundoplication.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Dybwik K, Tollai T, Waage Nielsen E, Brinchmann B.	Why does the provision of home mechanical ventilation vary so widely?	qualitative	2010 Chronic Resp Disease 7(2):67-73	3	6	6 focus groups from 5 hospitals in 5 counties in Norway involving nurses, medical specialties (n=34)	Group-based discussions until no new data or variables emerged on causes of HMV differences between counties.	N/A	N/A	To identify differences in treatment prevalence of HMV between Norwegian counties	Uneven distribution of “enthusiasm” involving high expertise and willingness, requirement for high competence, spreading competence and individual attitudes.	Norwegian Neuromuscular Diseases Foundation and Nordland Hospital.	HMV considered a new, highly specialized treatment for relatively few patients – importance of spreading competence with collaboration rather than reliance on the few unevenly distributed enthusiastic experts

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Eagle M, Baudouin S V, Chandler C, Giddings D R, Bullock R, Bushby K	Survival in Duchenne muscular dystrophy: improvements in life expectancy since 1967 and the impact of home nocturnal ventilation	Qualitative	2002 Neuromuscular Disorders	3	184	Patients with DMD attending single centre where NIV has been used since mid 1990's. 184 patients identified. 134 died from respiratory failure, 25 from cardiomyopathy. None were treated with steroids.	None - retrospective case note review	Survival with or without NIV	Retrospective case note review 1967-2002	Survival	Apparent decade on decade improved survival (assessed as decade of death) was all due to influence of 24 patients on NIV. Apart from improvement after 1960's, there is no apparent improvement in survival in the group without NIV with time. Of the 24 who have been started on NIV 15 are still alive. Excluding those that died of cardiomyopathy, mean survival in non-vent group was 19.29 years (n = 134, 95% confidence interval 18.61, 19.97) compared to a mean survival of 25.3 years for those who were ventilated (n= 24, 95% confidence interval 23.11, 26.58). 15 are still alive, so mean survival will increase. Chances of surviving to 25 years in ventilated group was 53% compared with 12% for those who died in 1980s without ventilation.	Not stated	Reasonable evidence that ventilation prolongs life in pts with DMD. Mention, but don't discuss in any detail cough assist or improved physio or ICU which might also affect outcome.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow -up	Outcomes	Effect size	Funding	Comments
Engel J, D Kartin, G.T Carter, M P Jensen, K . M Jaffe	Pain in youths with neuromuscular disease	qualitative	2009 Am J Hospice and Palliative medicine	3	42	Youths with NMD. 24 males (57%), 18 females (43%) age range 8-20 years. 14(37%) DMD commonest diagnostic group. 21(50%) ambulatory	To study prevalence and characteristics of pain in children and young people with NMD by questionnaire	Comparison of results with parental report	N/A	Pain frequency and rating scale, pain interference with life domains; parental report, level of motor function,	23(55%) youth experience chronic pain(>3 mths) Current pain intensity was 1.3 (range 0-6) mean intensity 2.39 and duration 8.75 hrs. Most commonly in legs. 83% pain medication	National Institute of Child Health and Human development/ National Institute on disability and rehabilitation research training centre	Pain is common and under reported. 30(71%) parents reported chronic pain vs 23(55%) youths. Note further factor of under reporting compared to parental report.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow -up	Outcomes	Effect size	Funding	Comments
Erby L, Rushton C, Geller G.	“My son is still walking”: Stages of receptivity to Discussions of Advance Care Planning Among Parents of Sons with Duchenne Muscular Dystrophy	7	2006 Semin Padiatr Neurol 13:132-140	3	17	Parents with a son with DMD aged 7+yrs recruited through invitation letters from US states and Muscular Dystrophy Association	Semistructured interview on Experiences of communication, planning and discussion of the future including knowledge of and experience of ACP	N/A	N/A	Common themes based upon topics, analysed by qualitative data software package	Parents were unfamiliar with ACP. None felt ready to discuss end of life decisions but all expressed a need for respite services. ACP communication influenced by competing demands, needing to distance themselves at times, absence of expert communication guidance	Not reported	Use of windows of opportunity help in particular discussions as sons approach transitional milestones

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Evans GA, Drennan JC, Russman BS	Functional classification and orthopaedic management of spinal muscular atrophy	Qualitative	1981 J Bone Joint Surg	3	54	Children with infantile and juvenile SMA reviewed ~17 years following diagnosis	None	Progression of weakness and limb or spinal deformity	Average of 17 years	Independent sitting, scoliosis, limb deformity	Forty seven of 54 children developed scoliosis. Eleven received scoliosis surgery. Scoliosis progressed more rapidly in wheelchair bound children. Use of bracing slowed the progression of scoliosis.	Not stated	A descriptive paper of practice in a single unit over many years.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Fauroux B, Lavis J-F, Nicot, F, Picard A, Boelle P-Y, Clement A, Vazquez M-P	Facial side effects during noninvasive positive pressure ventilation in children	Qualitative	2005 Intensive Care Med	3	40	All patients under care of a single centre on home nasal or face mask NIV or CPAP for at least 1 month. 16 had OSA, 14 had neuromuscular disease and 10 had CF.	Use of nasal mask for CPAP or BiPAP	None – observational study	Cross-sectional single time point assessment of facial side effects	Presence of facial side effects.	Skin injury was observed in 19 patients (48%). – mostly redness, and skin necrosis in 3 (8%). Global facial flattening was observed in 68% of the patients. Global facial flattening was not associated with age (OR=0.4, 95% CI 0.1–1.5, p=0.18), type of mask (OR=0.4, 95% CI 0.1–1.6, p=0.20), or daily use of NPPV (OR=1.8, 95% CI 0.4–7.4, p=0.46) Maxillary retrusion was observed in 37% . Daily use (>10 h per day) was associated with maxillary retrusion (OR 6.3, 95% CI 1.3–29.3, p=0.02).		Shows that there are common side effects from use of facial masks. Custom masks may prevent local skin damage. Facial flattening is harder to avoid. Not clear whether the side effects where present at the time of assessment or at any time during the use of the mask.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow -up	Outcomes	Effect size	Funding	Comments
Fauroux B, Leroux K, Desmarais G, Isabey D, Clément A, Lofaso F, Louis B	Performance of ventilators for noninvasive positive pressure ventilation in children	Qualitative	2008 Eur Respir J	3	N/A	N/A	N/A	Performance of 17 different ventilators	N/A	N/A	Bench test of 17 ventilators used for NIV in children in France. Twelve ventilators were pressure targeted ventilators, one ventilator was a volume targeted ventilator, and 4 ventilators had the 2 modes. None of the ventilators had appropriate insp or exp triggers for the infant simulation.	Not stated	Shows that most ventilators cannot synchronise with ventilators – inspiratory or expiratory

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Fauroux B. Guillemot N. Aubertin G. Nathan N. Labit A. Clement A. Lofaso F.	Physiologic benefits of mechanical insufflation-exsufflation in children with neuromuscular diseases	Qualitative research	2008 Chest	3	17	Children with NMW who's diagnosis included: DMD n=4, SMA n=4, merosin deficit CMD n=1, spina bifida n=1 congenital myopathy n=7. All in stable state and recruited from out patient clinic.	Effects and tolerance of mechanical insufflation-exsufflation at pressures of 15, 30 and 40 cmH_2O	Changes in airway pressure, PEF, SNIP, expiratory volume, breathing pattern, SpO2 and etCO2 with mechanical insufflation-exsufflation at pressures of 15, 30 and 40 cmH2O	Single Visit.	Primary outcome: airway pressure and airflow. Secondary outcomes: SNIP post intervention, Respiratory comfort, expiratory volume, breathing pattern, SpO2 and etCO2	The airway pressure measured on the facial mask during the mechanical insufflation-exsufflation applications were constantly lower than the inspiratory and expiratory pressures set on the device. A pressure-dependent increase in the maximal and mean inspiratory and expiratory flows was observed during the three MI-E applications). This translated into a proportional increase in Vexp during the MI-E manoeuvre Mechanical insufflation-exsufflation had no significant effect on breathing pattern SpO2 remained stable throughout. No adverse events reported.	BF was supported by the association Francaise contre les \myopathies, assistance publique-hospitaux de paris, Institut national se sante et de la recherche medicale m \leg poix and universati e Pierre et Marie Curie-Paris and ADEP assistance	Study shows that results confirm the good tolerance and physiologic short term benefit of the MI-E in children with neuromuscular disease who were in a stable state. In order to get the greatest change in expiratory airflow rates high inspiratory and expiratory pressures were required. The pressure record on the device is less than the pressure measured at the mouth

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Fraser L K, Aldridge J, Manning S, O'Leary S, Miller M, McCulloch R, Childs A M	Hospice provision and usage amongst young people with neuromuscular Disease in the Untied Kingdom	qualitative	2011 Eur J Paediatr Neurol	3	27	756 NM patients cared for by 27/31 UK children's hospices across UK England (22), Scotland (2), Wales (2) Northern Ireland (1)	Survey of UK hospices providing for NM patients during 2007-2008	N/A	N/A	Questionnaire on service usage and provision over study period 2007-2008. Hospice usage by 89 DMD patients at one specific centre.	756 patients comprised 17% total hospice population (5-35%). Referrals /yr n=5(SD 5) Only 7/47 (14.8%) died in the hospice compared with 134/531 (25.2%) with other diagnoses. Only 18(67%) hospices had psychological support., 17 (63%) had community services and 24 (89%) had medical support. 73% visits were planned stays	2 year Heartsongs Project Funding from Martin House Children's hospice, Leeds.	NM patients form a large proportion of hospice caseload. Many valued services are not NHS funded. Fewer adult services for onward referral

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow -up	Outcomes	Effect size	Funding	Comments
Galasko, C. S., J. B. Williams on and C. M. Delaney	Lung function in DMD	Qualitative	1995 Eur Spine J	3	181	181 DMD patients 93 pt accepted standing regimen 48 pt underwent spinal surgery; 28 who were fit for surgery refused	Standing regimen for patients who went off their feet Spinal stabilisation	Changes in lung function and scoliosis were compared between those stood and those who did not accept the standing regimen	Up to 8 years	Lung function	Pt who complied with standing regimen had a sig better lung function (FVC and PEFR) than those who did not stand. Spinal stabilisation slowed down the progression of scoliosis and prolonged survival. Pt who underwent spinal surgery had sig better lung function compared to those who refused surgery.	Not stated	Although the number of subjects involved is far more than some smaller case series, this is not a randomised controlled trial. The authors did point out the lung function between interventional and non-interventional groups were comparable at the start of the study. Lung function was reported in absolute values, rather than percent predicted, which may mask/under-estimate the true extent of the effect of age and intervention on lung function.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Gauld, L. M., J. Kappers, J. B. Carlin and C. F. Robertson	Prediction of childhood pulmonary function using ulna length	Cross sectional study	2003 Am J Resp Crit Care Med	2+	2343 subjects	Normal children of 5.3 to 19.6 years	Measurement of pulmonary function, height and various limb measurements	Between lung function calculations based on height and based on various limb parameters.	Not applicable	Ulna length predicts FVC and FEV1 in healthy children	Good correlation	Local research grant	Prediction equations for FEV1 and FVC derived from ulna length

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Gauld, L. M., J. Kappers, J. B. Carlin and C. F. Robertson	Height prediction from ulna length	Cross sectional study	2004 Dev Med Chil Neurol	2+	2343 subjects	Normal children of 5 to 19 years	Measurement of height, armspan, forearm, ulna, tibia and lower leg	Between different measures and the height	Not applicable	Ulna length predicts height in normal children	Not applicable	Not stated	Cross sectional centiles available. Ulna length superior to other measures in predicting height.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Gauld, L. M., A. Boynton, G. A. Betts and H. Johnston	Spirometry is affected by intelligence and behavior in Duchenne muscular dystrophy	Prospective study	2005 Ped Pulm	2+	Boys with DMD	Recruited at 6-19 years	Spirometry measured. Performance and verbal IQ, Behavioural scales.	Measurements with and without computer visual incentives.	Recruited over 12 month period	Spirometry is affected by intelligence and can be improved using visual incentives in those with moderate intellectual or behavioural impairment	Not applicable	Not stated	These techniques may allow more representative measures of lung function in those with intellectual impairment.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Gauld, L. M. and A. Boynton	Relationship between peak cough flow and spirometry in Duchenne muscular dystrophy	qualitatiive	2005 Pediatr Pulmonol	3	47 boys	Duchenne MD Mean 12.6 years, range 6.0 to 18.6 years	Spirometry and Peak Cough Flow (PCF) measured in each subject.	Between the 2 measures	Not applicable	VC < 2.1 litres predicts PCF of < 270 l/min. PCF correlates with FVC and PEF.	Not applicable	Not stated	Authors state that PCF is not measured in all centres and examined whether there was any correlation between conventional spirometry measures and PCF

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow -up	Outcomes	Effect size	Funding	Comments
Geesvasinga N, M.M.Ryan	Physician attitudes towards ventilator support for spinal muscular atrophy	Qualitative	2007 J of Paediatrics and Child Health	3	92	92/157 physicians- 16 geneticists, 19 intensivists, 28 neurologists, 29 respiratory physicians. All major paediatric hospitals in Australia and New Zealand	To survey attitudes towards ventilatory management of children with spinal muscular atrophy type 1.	Differing attitudes amongst specialties	N/A	Opinions to 16 to questions posed on NIV and invasive ventilator support	59% response rate. 47% opposed invasive ventilation for respiratory failure in SMA type 1. 85% felt NIV support for acute episodes and 49% for chronic resp failure. Most felt policy and ethic committee helpful where areas of disagreement	Not reported	Most opposed invasive support for SMA type 1 but consider NIV acceptable. Suggestion of guidance from hospital ethics or a policy where there is disagreement.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow -up	Outcomes	Effect size	Funding	Comments
Gibson B	Long-term Ventilation for Patients with Duchenne Muscular Dystrophy	qualitative	2001 Ethics in Cardiopulmonary medicine	3	45	Physicians of patients with DMD through Canadian neuromuscular clinics	To describe Long term ventilation (LTV) attitudes and practices of Canadian physicians	N/A	N/A	Mail questionnaire and semi-structured interviews	25% physicians do not discuss LTV Most frequently cited reason : poor patient Quality of life (52.6%)	Ontario Respiratory Society Fellowship	Demonstrates considerable agreement amongst physicians about disclosure practices which are often subjective with little family participation

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Gilgoff I, W Prentice, A Baydur	Patient and Family participation in the management of respiratory failure in Duchenne's Muscular Dystrophy	qualitative	1989 Chest	3	11	15 DMD patients mechanically ventilated, av age : 19 yrs, 7 mths .10 alive.	Description of clinic established for patients with impending respiratory failure – making informed choices	N/A	7 years	Description of clinic and patients decisions	Prediction of respiratory failure possible in 14/15. 11/15 electively chose ventilation. 2 chose natural death., 4 could not decide and 3 – choice not available	Not reported	Patients and families can be active participants in life and death decisions

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow -up	Outcomes	Effect size	Funding	Comments
Gill, I., M. Eagle, J. S. Mehta, M. J. Gibson, K. Bushby and R. Bullock	Correction of neuromuscular scoliosis in patient with pre-existing respiratory failure	Qualitative	2006 Spine	3	8	8 subjects (6M;2F) aged 8-15 yr All have underlying myopathies All needed nocturnal ventilation prior to spinal surgery	All underwent spinal surgery	Within subject comparison – pre and post op Cobb angle and FVC	12-80 months post op	ICU stay Hospital stay post op cobb angle FVC Complications	Difference between pre and post op Cobb angle was sig (p=0.0002) No sig difference between pre and post op FVC	Not stated	Authors concluded higher risk gp of NMD patients with resp failure can have a spinal surgery with no complications – which was attributed to prior correction of hypoventilation. Small case series

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow -up	Outcomes	Effect size	Funding	Comments
Goldstein , Meyer and Freund	Effects of overfeeding in Children with Muscle Dystrophies	qualitative	1989 J of Parenteral nutrition	3	10	6 Duchenne MD (aged 10 to 16 yrs) and 4 Congenital MD (10 to 18 years)	nightly additional feeds of 1000cals and 37.2g protein/subject/night. Anthropometry, lung function, muscle strength and 24 hr urinary excretion of creatinine (as a measure of muscle breakdown)		3 months		In the CMD group, the body weight did not change, staying at 50% ideal for age. In the DMD group, body weight increased from 31.6 (17.9-35.8)Kg to 35.8Kg (21.3 to 58.5) ($p<0.05$). In the DMD group, there was a 13% (9-14) increase in midarm circumference ($p<0.05$) and 9.2% increase (8.8 to 9.5) in triceps skin fold thickness. In the CMD group, no change occurred. Baseline nitrogen balance was mildly positive in both groups but significant in the DMD group ($p<0.05$) No change in either group in lung function		Nightly overfeeding contributed to improved body weight and relative muscle mass in DMD patients (but not this CMD group) but has not been shown by this study to improve lung function or muscle strength over this very short study period.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow -up	Outcomes	Effect size	Funding	Comments
Gomez-Merino E, Bach JR	Duchenne muscular dystrophy prolongation of life by noninvasive ventilation and mechanically assisted coughing	Qualitative	2002 Am J Phys Med Rehabil	3	91	Retrospective case note review of 125 patients with DMD referred to single tertiary centre from 1983 to 2002. 91 had used ventilatory support and were considered the study population. No patients had used steroids. 57 had NIV and manual assisted coughing. 34 had oximetry driven use of mechanical cough assist.	Effect on survival of the use of full-time noninvasive IPPV with and without access to protocol of using oximetry driven cough assist. Survival was considered to have been prolonged when patients needed >16 hours per day NIV.	Patients with and without 'access' to the protocol. Those without access are presumably those cared for before 1993 when the protocol was introduced	None - retrospective case note review	Survival. Cause of death subdivided into respiratory or cardiac	Of the 57 non-protocol patients, 31 died, 14 from resp failure and 6 following tracheostomy, 7 from heart failure, 1 suicide and 3 unknown. Of the 34 protocol patients, only had 3 died – all from heart failure. There were no respiratory deaths. Suggested 30 year survival at 30 years was 80% for protocol patients versus 45% in non-protocol. 14/57 in the non-protocol group had a tracheostomy versus 0/34 in the protocol group.	Not stated	Although there may be unknown confounders since this is an uncontrolled study using historical controls, it does suggest that NIV combined with oximetry driven mechanical cough assist protocols prolong life and avoid the need for tracheostomy at least in this centre.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Gonzalez, J., T. Sharshar, N. Hart, K. Chadda, J. C. Raphael and F. Lofaso	Air leaks during mechanical ventilation as a cause of persistent hypercapnia in neuromuscular disorders	Qualitative	2003 Intensive Care Medicine	3	95	All had neuromuscular weakness – 57 DMD (mean age 34), 21 myotonic dystrophy (mean age 61) 17 others (mean age 53). All used ventilation at least 6 hours per day. 43 nasal mask, 52 cuffless trachy.	Effects of leaks in ventilation on PaCO2	Arterial CO2 measured following a night of ventilation and immediately prior to being ventilated. Leak was calculated on the basis of a 2 limbed ventilator circuit in these patients all using volume cycled ventilators. Leak was assumed to be the difference between inspired and expired volumes. Leak was calculated over 10 consecutive breaths during wakefulness.	Study over a single night in patients seen for annual assessment	PaCO2	Average leak was 25% for NIV and 21% for trachy vent. Despite limitations in the method of leak assessment there was a weak correlation between leak and both night time and day time PaCO2. Of the 14 patients with hypercapnia (>45mmhg), 5 were improved with a chin-strap, 2 were switched to pressure support (one improved), 5 were recommended for trachy. Of those with trach already, CO2 was improved by increasing tidal volume or increasing size of trachy tube.	Not stated	Authors think that the leak related to poor mask fit, leak round trach or mouth leak. They don't think it reflects poor lung compliance since VC did not differ between hypercapnic and non-hypercapnic group. Nevertheless correlation between CO2 and leak was weak and only accounted for 8% of the variance of CO2. Leak assessment may not be accurate since leak can also occur during expiration; further more leak is greater during sleep with decreased muscle tone, and may be dependent on posture.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Gozal, D. Thiriet, P.	Respiratory muscle training in neuromuscular disease: long-term effects on strength and load perception	RCT	1999 Med Sci Sports Exerc	1-	21	Children with NMD	inspiratory and expiratory muscle training (threshold IMT™ and threshold PEP™)	Training versus no training Training thresholds were initially set at the lowest pressure (7cmH_2O) for 1 minute followed by a 1 minute rest period with increasing the pressure by 2 cmH_2O until they reached their targeted training pressure of at least 30% of their Pi_{max} or Pe_{max}.	Measurements were made 3 months prior to randomisation, commencement of the study, 3 and 6 months (end of training period) followed by 9, 12 and 18 months.	RMT was shown to have beneficial effects in terms of Pi_{max} and Pe_{max} in the trained group of patients with NMD for the duration of the training period. At the cessation of training Pi_{max} declined towards the baseline value.	Training in neuromuscular disorder patients was associated with improvements in Pi_{max} (mean delta max: +19.8 +/- 3.8 cmH2O in trained vs +4.2 +/- 3.6 cmH2O in non trained; $P < 0.02$) and in Pe(max) (mean delta max: +27.1 +/- 4.9 cmH2O in trained vs -1.8 +/- 3.4 cmH2O in non trained; $P < 0.004$). Similarly, respiratory load perception significantly decreased during the RMT period in trained (mean delta: 1.9 +/- 0.3; $P < 0.01$) but did not change in non trained (-0.2 +/- 0.2)	DG received various grants none direct for this study	RMT improved Pi_{max} and Pe_{max} for the training period. Patients also reported that they were less breathless as rated on the BORG score

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Jeppeson J., A. Madsen, J. Marquardt, J. Rahbek	Living and ageing with spinal muscular atrophy type 2: Observations among an unexplored patient population	Qualitative	2010 Develop. Neurorehab.	3	29	All SMA type 2, registered with the RCFM. 17 men aged 18-63, 12 females aged 18-69yrs.	To study Conditions of living, participation and diagnostic perceptions in a national population of adults with SMA2	Some with general population with respect to marital status, employment, finances and education	N/A	Questionnaire, interview and narrative inquiry	Most stated their quality of life was fine. 90% always or most often had lots of energy though 1/3 had pain daily or weekly. Half answered that they worried about their disease and also the future (physical consequences of the disease upon their lives). Both men and women participated actively and frequently in everyday activities by deciding what has to be done.	Not reported	Adults with SMA 2 here played active roles in the direction of their own lives even though they were heavily dependent on others. Author touches upon the importance of the disability organisational networks which help to support parents and families to maximize independence.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow -up	Outcomes	Effect size	Funding	Comments
Hardart M.K, Jeffrey, Burns, Truog	Respiratory support in SMA Type 1 : A survey of physician practices and Attitudes	Qualitative	2002 Paediatrics	2	187	75/132 intensivists 61/155 physiatrists 51/150 paed neurologists from National US Societies who care for SMA type 1 patients	to study variability in practice of physicians treating respiratory failure in children with SMA 1	N/A	N/A	Questions following a scenario of a patient with SMA1 who was in respiratory distress - attitudes to questions based upon a Likert scale	Overall NIMV offered and recommended in 70% offered but not recommended in 22.5%, neither offered or recommended in 7.5% Significant differences between specialties -	not reported	wide variation in physician practices regarding mechanical ventilation of patients with SMA1 and therefore what would be recommended.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Harper CM, Ambler G, Edge G	The prognostic value of pre-operative predicted forced vital capacity in corrective spinal surgery for Duchenne's muscular dystrophy	Retrospective cohort study	2004 Anaesthesia	3	45	Consecutive patients with DMD undergoing scoliosis surgery in a single unit	Scoliosis surgery	Lung function (FVC) was assessed prior to planned surgery	Hospital discharge	The outcome of surgery (as assessed by duration of surgery, blood loss, duration of intubation and length of stay)	No outcome difference was found between those patients whose FVC was above or below 30% predicted	Not stated	20 of the 45 patients had FVC <30% predicted. No differences in characteristics between the groups was evident. There were more complications (25% vs 16%) in the patients with a lower FVC. The authors emphasise the importance on NIV to facilitate extubation.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Hibbert M.E., Lanigan A., Raven J., Phelan P.D.	Relation of armspan to height and the prediction of lung function	Qualitative	1988 Thorax	3	512 children	Healthy individuals recruited either at 8 or 12 years of age	Annual measurement of armspan and height	Relation of each measure to the other over late childhood growth	6 years	Prediction equations are available that allow a prediction of height from armspan measurement .	Not applicable	Not stated	These data allow estimation of height in non-ambulant children ion whom height measurement may be difficult or inaccurate.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow -up	Outcomes	Effect size	Funding	Comments
Hill M, Hughes T, Milford C	Treatment for swallowing difficulties (dysphagia) in chronic muscle disease	Qualitative	2004 Cochrane Collaboration	3	nil	Studies on adults and children with NMD investigating dietary intervention for dysphagia		N/A	N/A	Nil met inclusion criteria		Nil reported	Single observational study suggests that enteral feeding via gastrostomy is beneficial in congenital myopathy where dietary and swallowing advice alone did not appear effective.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow -up	Outcomes	Effect size	Funding	Comments
Hill M, Phillips	Service provision for adults with long-term disability : A review of services for adults with chronic neuromuscular conditions in the United Kingdom	Qualitative	2006 Neuromuscular Disorders 16:107-112	3	30	30 clinicians from 24/32 Muscle clinics in 25 UK cities	Questionnaire to determine level of service provision	N/A	N/A	Descriptors only	42% had services for transfer to adult care. 32% NMD clinics didn't routinely monitor resp function. 10% did not routinely undertake ECG. Fully funded domiciliary resp support in 43%. Wait for wheelchairs >6 mths : 45% and 73% cases. Inconsistent access to nursing and therapy services	work on behalf of the Muscular Dystrophy campaign	Only 11/24 (42%) had services to transfer towards adult care excluding those teams seeing both. Poor Local knowledge. A paucity of services for adolescents and young adults

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow -up	Outcomes	Effect size	Funding	Comments
Hodges L, B Dibb	Social Compariso n within Self-help gropus. Views of parents of children with Duchenne Muscular Dystrophy	qualitative	2010 J of Health Psychology 15(4); 483-492	3	8	Parents of children with DMD (7 mothers, 1 father) sons' ages ranged from 2yrs 7 months to 26 yrs. Time since Diagnosis 1 yr to 23 yrs	Qualitative approach to examine experiences of participation in family support group for DMD, interviews conducted	N/A	N/A	Semi structured interviews to identify common themes	Several themes emerged –around social comparison, including upward and downward comparison and strategies used to coping dimensions, with both positive and negative affects.	None reported	Coping through social comparison may have very different consequences for coping with problems that are essentially unchangeable than for problems that are more controllable

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Hukins, C. A. and D. R. Hillman	Daytime predictors of sleep hypoventilation in Duchenne muscular dystrophy	Cross sectional study	2000 Am J Resp Crit Care Med	2-	19 subjects from 28 potential recruits	Duchenne MD, all 12 years or older	Respiratory function measurements and polysomnography (PSG)	Between daytime lung function measures and PSG findings	None	PaCO2 of more than 45 mmHg predicts total sleep time below 90% oxygen saturation.	Not applicable	Not stated	Authors suggest that FEV1 below 40& predicted should be a trigger for assessing blood gases.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow -up	Outcomes	Effect size	Funding	Comments
Inal-Ince, D., S. Savci, H. Arikan, M. Saglam, N. Vardar-Yagli, M. Bosnak-Guclu and D. Dogru	Effects of scoliosis on respiratory muscle strength in patinets with neuromuscular disorders	Qualitative	2009 The Spine Journal	2-	63	22 NMD with scoliosis 17 NMD pt with no scoliosis 24 age and sex matched healthy controls	none	To compare lung function and respiratory muscle strength between groups prospectively	Not stated	Lung function including FVC, FEV1,PEFR. Oxygen saturation Respiratory muscle strength: MIP and MEP	NMD patients (with or without scoliosis) had sig lower PEFR, MIP, MEP than those of healthy subjects ($p<0.5$) NMD pt with scoliosis had sig lower values for FVC, FEV1 than those with NMD but without scoliosis and controls ($p<0.50$)	Not stated	Heterogeneity of NMD patients may have implication on the progression of the disease, scoliosis and consequently lung function Between scoliotic and non-scoliotic NMD pt, known confounding factors including age, duration of illness are unequally distributed across the groups

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Ishikawa Y. Bach JR. Komaroff E., Miura T. Jackson-Parekh R.	Cough augmentation in Duchene muscular dystrophy	Qualitative research	2008 Am. J. Phys. Med. Rehabil.	3	63 and 61 patients results were included in the study	Patients with DMD VC range (170-1980)ml who were residents or outpatients of a long term care facility in Japan. All patients were wheelchair dependent , ages range 12-36 years. 23 did not require NIV. 14 used nocturnal NIV, (10= volume controlled, 4= pressure controlled). Remainder n= 20 full NIV for up to 14 years.	Patients underwent the following cough augmentation methods in a random order: Manual assisted cough, Maximum insufflation capacity, Combination of maximum insufflation capacity with a manual assisted cough	Baseline unassisted PCF to PCF with Manual assisted cough, Maximum insufflation capacity, Combination of maximum insufflation capacity with a manual assisted cough	One off assessment	Effect of Manual assisted cough, Maximum insufflation capacity, Combination of maximum insufflation capacity with a manual assisted cough On PCF	Baseline unassisted PCF 138 ± 70L/min Manual assisted PCF 204 ± 75L/min Maximum insufflation capacity 236 ± 68L/min Combination of maximum insufflation capacity and manual assisted cough 302 ± 78L/min significant increase from baseline unassisted PCF with each cough augmentation technique ($p<0.0001$) there was also a significant difference between each intervention ($p<0.0001$)	Not funded.	Maximum insufflation capacity before a Manual assisted cough is more important than either alone. Maximum insufflation capacity easy to learn. Manual assisted cough is easily taught to carers. Maxim insufflation capacities can be achieved with a volume cycled ventilator and not pre set pressure cycled ventilator. Greatest improvements in PCF were seen inpatients with weaker coughs

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow -up	Outcomes	Effect size	Funding	Comments
Jenkins, J. G., D. Bohn, J. F. Edmonds, H. Levison and G. A. Barker	Evaluation of pulmonary function in muscular dystrophy patients requiring spinal surgery	Qualitative	1982 Critical care medicine	3	48	39 children with DMD, 6 with SMA and 3 others, who underwent 50 operations	Spinal surgery including Harrington procedure, Luque instrumentation and Dwyer procedures	Comparing rate of decline in VC (pre op and post op VC) between the operated patients and un-operated DMD patients	Not clearly stated	Lung function Days of ventilation post op Length of stay in ICU	Spinal fixation did not arrest the decline in lung function in patients but did slow down the rate of deterioration compared to pre-op changes (20%/yr vs 5% /yr)	Not stated	There is no clear explanation how and why 11 operated pt with DMD were chosen to compare with 31 non-operated DMD patients who were used as controls or if some of the pre-op data included the pre-op lung function of operated patients. There is no clear explanation how and why 11 operated pt with DMD were chosen to compare with 31 non-operated DMD patients who were used as controls or if some of the pre-op data included the pre-op lung function of operated patients.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Jeppesen J, Green A, Steffensen B F, Rahbek J	The Duchenne muscular dystrophy population in Denmark, 1977–2001: prevalence, incidence and survival in relation to the introduction of ventilator use	Qualitative	2003 Neuromuscular disorders	3	243	All patients with DMD in Denmark are cared for in 2 specialist centres. Data from these centres was used to calculate survival and to see of there was an association with the use of home ventilation. Prior to 1990s home ventilation for DMD was not standard practice in Denmark; since then NIV is introduced when needed and switch to trachy once daytime ventilation required.	Home ventilation	Survival over time	Retrospective review of survival	Survival	Twenty years' survival increased from 41.3% (95% CL: 24.2–58.6) for DMD patients diagnosed 1972–1976 to 66.6% (95% CL: 52.1–81.1) for DMD patients diagnosed 1977–1981. The overall prevalence proportion of mechanical ventilator users per 100 DMD patients increased from 0.9 (95% CL: 0–2.6) in 1988 to 43.4 (95% CL: 35.4–51.5) in 2002	Muskelsvindfonden and the Institute of Neuromuscular Diseases	Survival of Duchenne muscular dystrophy patients has increased and ventilator use is probably the main reason

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Jeppeson J., A. Madsen, J. Marquardt, J. Rahbek	Living and ageing with spinal muscular atrophy type 2: Observations among an unexplored patient population	Qualitative	2010 Develop. Neurorehab.	3	29	All SMA type 2, registered with the RCFM. 17 men aged 18-63, 12 females aged 18-69yrs.	To study Conditions of living, participation and diagnostic perceptions in a national population of adults with SMA2. Semi-structured questionnaires and interviews (n=29), in-depth interviews and personal narratives(n=3)	Some with general population with respect to marital status, employment, finances and education	N/A	Questionnaire, interview and narrative inquiry. Information on socioeconomic data, activities and personal significance of problems	Most stated their quality of life was fine. 90% always or most often had lots of energy though 1/3 had pain daily or weekly. Half answered that they worried about their disease and also the future (physical consequences of the disease upon their lives). Both men and women participated actively and frequently in everyday activities by deciding what has to be done. All had government assisted care (83% for 24 hrs).. Difficulties with fingers, increased with age. 50% worried about disease progression and the future.	Not reported	Adults with SMA 2 here played active roles in the direction of their own lives even though they were heavily dependent on others. Author touches upon the importance of the disability organisational networks which help to support parents and families to maximize independence. Deterioration of physical abilities was a major concern for women.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Johnson B.E., Westgate H.D.	Methods of predicting vital capacity in patients with thoracic scoliosis	qualitative	1970 J Bone Joint Surg Am	3	139 healthy subjects, 75 with thoracic scoliosis	Healthy subjects were 6 to 30 years old. Most of those with scoliosis were in the second decade.	Measurement of armspan, tibial length and height in normal. Armspan, height, Cobb;s angle and VC assessed in those with scoliosis	Prediction of height from armspan and tibial measures in the normal subjects. This was then compared to height predication using existing methods based on Cobb's angle in those with scoliosis.	Not applicable	Height can be predicted from limb measures in those with scoliosis.	Not applicable	Not stated	The authors are keen to point out that estimation of lung function from limb measures is superior to calculations using spinal curvature for 2 reasons: firstly that estimates using spinal curve lose correlation with worsening curve and secondly that using limb measures removes the need to have a recent spinal X-ray.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow -up	Outcomes	Effect size	Funding	Comments
Kang, SW, Bach JR.	Maximum Insufflation Capacity	Qualitative research	2000 Chest	3	43	DMD n= 9 (ages 22.5 ± 5.3), ALS n=6 (ages 53.9 ± 11.6), SMA n=3 (ages 11.3 ± 6.1), post polio n= 6 (ages 61.1 ±10.2), miscellaneous n= 6 (33.1 ± 20.7) years.	Twice daily 10-15 deep lung insufflations to maximum insufflation capacity in patients with neuro muscular disease who had a VC<2000ml.	1) Baseline VC and PCF compared to results post programme of maximum insufflation capacity twice daily 2) Comparisons made between 2 groups patients one with increasing maximum insufflation capacity and patients with a declining maximum insufflation capacity	6 months to am maximum of > 42 months	Changes in VC, maximum insufflation capacity and unassisted PCF and assisted PCF (maximum insufflation capacity and a manually assisted cough)	Maximum insufflation capacity increased from (mean ± SD) 1402 ± 530ml to 1711± 599ml ($p<0.001$) for 30 patients and decreased for 13. In those with increased maximum insufflation capacity an increase in assisted PCF from 3.7±1.4L/s to 4.3 ± 1.6L/s ($p<0.05$)	Not reported	With training the capacity to stack air to deep insufflations can improve despite progressive neuromuscular disease. This can result in increased cough effectiveness.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow -up	Outcomes	Effect size	Funding	Comments
Kang SW. Kang YS. Moon J H. Yoo TW.	Assisted cough and pulmonary compliance in patients with Duchene muscular dystrophy	Qualitative research	2005 Yosei Medical Journal	3	71	Males with DMD, mean age 14 ± 5.4 years old. All wheelchair dependant at 10.3 ± 1.8 years old.	Patients underwent the following cough augmentation methods: Manual assisted cough, Maximum insufflation capacity, Combination of maximum insufflation capacity with a manual assisted cough.	Baseline unassisted PCF to PCF with Manual assisted cough, Maximum insufflation capacity, Combination of maximum insufflation capacity with a manual assisted cough in 51 patients that could comply.	One off visit	Effect of Manual assisted cough, Maximum insufflation capacity, Combinatio n of maximum insufflation capacity with a manual assisted cough On PCF. Relationshi p between maximum insufflation capacity and VC	Baseline unassisted PCF 217.7± 65.9L/min Manual assisted PCF 250.6 ± 66.2L/min Maximum insufflation capacity 257 ± 65.8L/min Combination of maximum insufflation capacity and manual assisted cough 285.8 ± 75.9L/min A significant increase from baseline unassisted PCF with each cough augmentation technique ($p<0.01$). The difference between maximum insufflation capacity and VC was 438.4 ± 277.1cc	Not reported	Helping the weakened expiratory muscles whilst also increasing pre cough volume should be emphasized to optimally assist a cough in DMD patients and the study highlights that pulmonary compliance plays a crucial role in increasing an assisted PCF.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow -up	Outcomes	Effect size	Funding	Comments
Kang SW Kang Y, Sohn H, Park J, Moon J.	Respiratory muscle strength and cough capacity on patients with Duchene muscular dystrophy.	Qualitative research	2006 Yonsei Medical Journal	3	32	DMD patients Mean age 17.6 ± 5.1 years.	1) Maximum insufflation capacity. 2) manual assisted cough 3) combination of maximum insufflation capacity and manual assisted cough	Effect on PCF of the following interventions: 1) Maximum insufflation capacity. 2) manual assisted cough 3) combination of maximum insufflation capacity and manual assisted cough	Single visit	VC PCF and relationship between PImax and PCF	Baseline VC 1474 ± 632ml. Maximum insufflation capacity 1918 ± 586ml ($p<0.001$) Baseline PCF: 212±52L/min Maximum insufflation capacity PCF: 252 ± 45L/min Manually assisted cough PCF: 246 ± 49L/min Combined maximum insufflation capacity and manual assisted cough PCF: 286 ±41 L/min All 3 assisted cough techniques significantly increased PCF compared to baseline ($p<0.001$)	Not reported	All assisted cough techniques higher value than unassisted. Combined assisted cough technique significantly exceeded

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Katz S, Selvadurai H, Keilty K, Mitchell M, MacLusky I	Outcome of non-invasive positive pressure ventilation in paediatric neuromuscular disease	qualitative	2004 Arch Dis Child	3	15	Children with neuromuscular disease (mean age 11.7) eligible for NIV according to Toronto unit criteria: Either (a) evidence of sleep related hypercapnia ($PaCO_2$ greater than 50 mmHg) with desaturations to below 92% (n=10) or (b) a history of recurrent hospitalisations for pneumonia and/or atelectasis (n=5).	Effect of nocturnal NIV	Rate of hospitalisation and symptoms before and after NIV.	Hosptilisation rate for 1 year before and 1 year after NIV	Rate of hospitalisation and symptoms before and after NIV. Symptoms assessed prosepectively using questionnaire	Children spent 85% fewer days in hospital (mean pre-NPPV 48.0 days, mean post-NPPV 7.0 days, p,0.001) and 68% less days in intensive care after initiation of NPPV (mean pre-NPPV 12.0 days, mean post-NPPV 3.9 days, p,0.015) after initiation of NPPV. Of 7 school age children who completed questionairre, symptoms were better in 2.	Not stated	This study provides reasonable evidence that NIV can reduce hospital admissions in children with weakness. As the authors point out, young children may not be able to describe the symptoms of hypoventilation

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Katz S.L., Gaboury I., Keilty K., Banwell B., Vajsar J., Anderson P., Ni A., MacLusky I.	Nocturnal hypoventilation: predictors and outcomes in childhood progressive neuromuscular disease	Prospective cohort study	2010 Arch Dis Child	2+	46 children (6-17yrs)	Heterogenous group, none with DMD.	Polysomnography Pulmonary function, manual muscle strength, QoL measures	Presence of nocturnal hypoventilation assessed against measures assessed.	One year	Scoliosis predicts nocturnal hypoventilation, sensitivity 85%, specificity 73%.	See outcomes	Local research Foundations	Symptoms not a good predictor of nocturnal hypoventilation

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Keating J, Collins N, Bush A, Chatwin M.	High Frequency Chest Wall Oscillation in a Non-invasive Ventilation Dependent Patient with Spinal Muscular Atrophy Type I	Qualitative	2011 Respiratory care	3	1	SMA Type 1 severe ventilatory dependence	Addition of HFCWO	Pre post treatment chest x-ray and ventilator free time	One off hospital admission	Resolution of chest x-ray collapse consolidation	Single case	Not funded	No adverse events seen with the addition of HFCWO to standard treatment

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow -up	Outcomes	Effect size	Funding	Comments
Kennedy JD, Staples AJ, Brook PD, Parsons DW, Sutherland AD, Martin AJ, Stern LM, Foster BK	Effect of spinal surgery on lung function in Duchenne muscular dystrophy	Retrospective cohort	1995 Thorax	3	38	All patients had DMD	The study compared 17 who had scoliosis surgery and 21 who did not	Survival and rate of deterioration of lung function following surgery	7 years	FVC	FVC fell by 3.2% per year in the non-operated group and 5.4% per year in the operated group (NS). Survival was the same in each group.	Not stated	The study addressed some of the confounding factors in assessing changes in FVC over time. Pre-operative and post-operative rates of decline were the same.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Kennesoon A, J Kay Bobo	The effect of caregiving on women in families with Duchenne/ Becker Muscular Dystrophy	qualitative	2010 Health and Social Care in the Community 18(5), 520-528	3	1238	1238 women caring for individuals with DMD (1093)or BMD (140), non specified 5. Age 22-80 yrs (mean :43yrs) across US. Other variables – number cared for, state of ambulation	Questionnaires studying life satisfaction, stress and distress	Compared with data from BRFSS 2007 survey for prevalence of distress and marital status, age adjusted to US 2000 population for women>19yrs with children<18 yrs at home.	N/A	Zarit Burden Interview Kessler psychological distress scale ENRICHD Social Support Instrument	High level of caregiving demands reported by 50.4% based on ZBI. Ambulatory care recipients associated with low level ZBI (aOR=3.84,p<0.001) and high social support (aOR=3.16,p<0.001) High stress in 46.4%, serious psychological stress in 11.6% (general population 7.71%)	Supported by centers for Disease Control and prevention Battelle centers for Public Health reaserach and Evaluation, Seattle	High level of care demands reported 50.4% caregivers. Post ambulatory phase associated with decreased social support, increased ZBI scores. Distress and high stress predicted by low resiliency, low social support and low income.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Khan Y, Heckmatt J	Obstructive sleep apneas in Duchenne muscular dystrophy	Qualitative	1994 Thorax	2	21	21 non-ambulant DMD patients attending tertiary UK unit	Case control study of home polysomnography looking for sleep related breathing abnormality	12 age matched normal male subjects as controls (av age 14 yrs (10-22yrs)	10 had repeat sleep studies, 2 followed for 3 years	Polysomnography over 2 consecutive nights Questionnaire regarding symptoms	13/21 had O2sats<90% during sleep. With 12/13 patients : hypoxaemic apneas, most during REM sleep. Mean 60% all apnoeas (10-92%) were obstructive in nature, mixed in 10% and central in 30%. All normal subjects had few apnoeas but none below 90%O2 sat.	None declared	Hypoxaemic periods became more frequent at followup. In 2 subjects, obstructive apneas fell from 95% to 20% and 60% to 30%, but increase in hypoxaemic apneas occurred.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Kinali M, A ,Manzur, E ,Mercuri, B Gibson, L ,Hartley, A Smonds, F ,Muntoni,	UK Physicians attitudes and practices in long-term non-invasive ventilation of Duchenne Muscular Dystrophy	qualitative	2006 Pediatric Rehab 9(4):351-364	3	38	UK physicians identified through the Muscular Dystrophy campaign.27/38 (71%)paediatric, 5 adult or mixed service(13.2%), paediatric respiratory clinic (2.6%)	To survey attitudes of UK physicians to long-term Noninvasive ventilation	N/A	N/A	Modified version of validated Canadian questionnaire	81% felt ethically obliged to discuss NIV with families whilst 13% believed it offered a poor quality of life. 47% discuss it in-depth when in resp failure.		Different practices of disclosure of life prolonging ventilation options by different physicians. 71% wished for national consensus guidelines

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow -up	Outcomes	Effect size	Funding	Comments
Kinali M, Messina S, Mercuri E, Lehovsky J, Edge G, Manzur AY, Muntoni F	Management of scoliosis in Duchenne muscular dystrophy: a large 10-year retrospective study	Cohort study	2006 Developmental Medicine and Child Neurology	3	123	Adults (over 17 years) with DMD. Seventy had a scoliosis over 50^{o} and 43 of these went on to surgery.	None	The paper describes the characteristics of scoliosis in their practice of young adults	Surgical group 3m - 5.5yr Non-surgical group 2m -9y	Serial lung function Progression of scoliosis Cardiac impairment Subjective perception of comfort in sitting Survival Complications related to surgery	Of their patients, 10% had no scoliosis, 13% had a scoliosis between 0 and 30^{o}, and 13% between 30 ^{o} and 50^{o}. Surgery was only offered to those with a scoliosis> 50^{o}.	Not stated	The paper is a descriptive report of experience at a single unit.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Kirk, V. G., W. W. Flemons, C. Adams, K. P. Rimmer and M. D. Montgomery	Sleep-disordered breathing in Duchenne muscular dystrophy: a preliminary study of the role of portable monitoring	Observational study	2000 Pediatric pulmonology	3	11 boys with DMD	Aged 9-21 years	Measurements of sleep breathing parameters using portable home monitoring with Snoresat or Edentec devices.	Between daytime lung function, functional scores and sleep study findings	None	Portable home monitoring of oximetry can be performed at home	Not applicable	None stated	Three of the patients had sleep disordered breathing, all of whom had greatly reduced daytime lung function.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Koessler, W. Wanke, T. Winkler, G. Nader, A. Toifl, K. Kurz, H. Zwick, H.	2 Years' experience with inspiratory muscle training in patients with neuromuscular disorders	Qualitative	2001 Cheat	3	27	Patients with NMD (Duchenne muscular dystrophy, 18 patients; spinal muscular atrophy, 9 other) mean (± SD) age 16.30 ± 4.84 years old	Patients underwent a twice daily visual feedback strength training programme (of 10 breaths) at 70-80% of their Pi_{max}.	Vital capacity (. Maximal inspiratory pressure PI_{max} maximum voluntary ventilation test Ventilatory function and inspiratory muscle function were evaluated 6 months prior to training and every 3 months to the end of training at 24 months.	Patients were assessed 6 months prior to training and then followed up for a 2 year period	For all 3 groups there was a significant improvement of Pi_{max} ($p<0.007$) until month 10 when there was a plateau. There was also a significant improvement in MVV until the month 10 ($p<0.015$). There was no significant change or more importantly no decline in VC.	PImax values improved in group A (VC, 27 to 50% predicted) from 51.45 to 87.00 cm H_2O, in group B (VC, 51 to 70% predicted) from 59.38 to 94.4 cm H_2O, and in group C (VC, 71 to 96% predicted) from 71.25 to 99.00 cm H_2O. 12sMVV values improved in group A from 52.69 to 69.50 L/min, in group B from 53.18 to 62.40 L/min, and in group C from 59.48 to 70.5 L/min. Significant improvement of PImax ($p < 0.007$) and 12sMVV ($p < 0.015$) until the 10th month when a plateau phase was reached with no decline in the following month until the end of training.	Unknown	In stable state NMD patients without signs of nocturnal hypoventilation improvements are seen in Pi_{max} up to 10 months of training. Improvements in Pi_{max} are sustained with continued training (at least for 2 years as in the present study)

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Kohler M., C.F.Clarenbach, L. Boni, T. Brack, E.W.Russi and K.E.Bloch	Quality of life, physical disability and respiratory impairment in Duchenne muscular dystrophy	Qualitative	2005 Am J of Resp & Critical Care Med.	3	35	Males with DMD 18 living at home,17 in fulltime institution/school scoliosis in 34/35operated (n=24) Cardiomyopathy (n=10) NIPPV 14, 4 nearly 24 hrs.	Short form-36, HR-QuOL questionnaire, ADL score to 10 giving overall dependency score (max value 80)	To compare results of ventilated with those non-ventilated and ref values with US/German population	2 assessments separated by 1 or more years.	Short form36HRQUOL ADL rated Spirometry Cardiography Body weight/length.	In spite of low recorded physical functioning, general and mental health, social functioning, emotions and pain were not impaired, corresponding to normal population values.	Lung League of Zurich.	In spite of greater limitations in lung function and ADL, patients on NIPPV rated HRQUOL similar to those not on NIPPV.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Kotterba, S., T. Patzold, J.-P. Malin, M. Orth and K. Rasche	Respiratory monitoring in neuromuscular disease--capnography as an additional tool?	Observational study	2001 Clin Neurol Neurosurg	2-	11 patients	Slowly progressive neuromuscular disease Ages 15-75 years. One adolecscent. 6 with various Muscular Dystrophies, 3 with SMA.	None	Oximetry with end tidal CO2	Not applicable	All subjects with significant hypercapnoea also had evidence of oxygen desaturation	Not applicable	Not stated	This study suggests that if oxygen saturation is maintained then significant hypercapnoea will not be present

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Kotwicki T, Jozwiak M	Conservative management of neuromuscular scoliosis: personal experience and review of literature	Qualitative	2008 Disability & Rehabilitation	3	45	A descriptive review of nearly 1500 children with scoliosis, but only 45 of these had flaccid scoliosis (caused by SMA or myelomeningocele)	Conservative management (bracing)	Progression of scoliosis	Not stated	Progression of spine curvature (qualitatively described; no actual data)	22 of 45 patients had the progression of their scoliosis halted by using a suspension trunk orthosis (a brace that rests against the seat, not the pelvis).	Not stated	This is a descriptive paper with very little data. No methodology is described

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Kurz, L. T., S. J. Mubarak, P. Schultz, S. M. Park and J. Leach	Correlation of scoliosis and pulmonary function in DMD	Qualitative	1983 J of Ped orthopaedics	3	25	25 DMD patients	nil	Lung function of 25 DMD patients were correlated with age and degree of scoliosis	20 years of practice in a single clinic	Pulmonary function namely FVC, PEFR Spinal curvature	Age and degree of thoracic scoliosis are better predictors of FVC% than either variable alone Thoracolumbar curves are insig in adversely affecting FVC%	Not stated	Retrospective and longitudinal study

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Linderholm H., Lindgren U.	Prediction of spirometric values in patients with scoliosis	qualitative	1978 Acta Orthop Scand	3	209 normal subjects	91 males, 118 females, of 5-78 years	Measurement of height and of armspan. Spirometry.	Linear regression equations generated to describe the relation of the 2 measurements to spirometry	Further assessment of the effect of using armspan to predict height versus existing methods of height prediction in scoliosis using curvature.	Armspan used in a regression equation with age is a good predictoir of spirometry.	Not applicable	Not stated	These authors sought to prove that existing methods in 1978 that used curvature to “re-calculate” height for spirometry prediction were inferior to using armspan.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Evaluation of carbon dioxide rebreathing during pressure support ventilation with airway management system (BiPAP) devices	Lofaso F, Brochard L, Touchard D, Hang T, Harf M, Isabey D	Qualitative	1995 Chest	3	7, plus the use of a lung model.	Ventilated adult patients on ICU in stable respiratory failure; 3 had tracheostomies, 4 were intubated	Potential effect of re-breathing via BiPAP	Tidal volumes, work of breathing, blood gases with standard ICU ventilator, standard BiPAP or BiPAP with non-rebreathe expiratory valve, used in random order	All observations made on a single day.	Work of breathing, blood gases and tidal volume.	Using the model lung system with minimal PEEP (1.3cm) the residual volume of expired air in the system was 400ml (55% of the tidal volume). Adding the valve increased the PEEP and the expiratory work load by 50%. In the clinical study use of unmodified BiPAP without external PEEP increased WOB by 50% resulting in bigger tidal volumes (RR not changed). CO2 was not changed.	Not stated	CO2 re-breathing is possible using pressure-targeted ventilators if there is no external PEEP. Easiest way to avoid this is to use PEEP; also if there is a leak in the circuit, re-breathing is also likely to be minimal.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Mah J.K, J.E.Thannhauser, H. Kolski, D. Dewey	Parental stress and quality of life in children with neuromuscular disease	qualitative	2008 Pediatric Neurol.	3	109 families	19 had a child requiring home mechanical ventilation. More common diagnoses : DMD and Becker MD (22%), CMT(22%), SMA(16%). 75(69%) male. Mean age : 10.5+/-5.2 yrs	To study health-related quality of life and parental stress in those with NM disease who were ventilated and non-ventilated	Comparison of scored from non-ventilated children and those ventilated	N/A	Pediatric Quol of inventory Physical and psychosocial health summary Parenting stress index	Ventilated children had lower PedQUOL than non-ventilated (47.9vs 61.5 p=0.013) However parents did not report significantly more stress in caring for ventilated children.	Alberta Children's Hospital Foundation	Despite lower quality of life, parents did not consider caring for their ventilated children as more stressful than prior to ventilation – perhaps a consequence of adjustment over time.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Markström A, Sundell K, Lysdahl M, Anderssón G, Schedin U, Klang B.	Quality-of-life evaluation of patients with neuromuscular and skeletal diseases treated with noninvasive and invasive home mechanical ventilation.	Qualitative	2002 Chest	3	91	91 of 120 consecutive adult (mean age 59 yrs) patients from single centre responded to questionnaire. 60 with NIV. 33 post-polio, 16 had neuromuscular disorder, 13 scoliosis, and 29 had other diseases.	3 questionnaires: Sickness Impact Profile (SIP) - health-related dysfunction of daily living, the Health Index (HI) – perceived health status, and the Sense of Coherence (SOC) scale- comprehensibility, manageability, and meaningfulness in stressful life situations	Impact of home ventilation on QoL, comparing NIV to tracheostomy	Single questionnaire study	Questionnaire responses.	The quality of life scores, using 2 out 3 different measures, were not different between the NIV and tracheostomy groups. For the 3rd measure of quality of life there was significantly better score in the tracheostomy group, but the difference between the groups was small (NIV group score 25.2 +/- 3.6 compared to 27.8 +/- 3.7 in the tracheostomy group; possible scores range from 9 to 36).	Not stated	Adult patients with stable respiratory failure report good quality of life with either NIV or tracheostomy, and some patients prefer trachy.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Marsh A, Edge G, Lehovsky J	Spinal fusion in patients with Duchenne's muscular dystrophy and a low forced vital capacity	Retrospective cohort study	2003 European Spine Journal	3	30	Patients with DMD undergoing posterior spinal fusion . 17 had a pre-operative FVC > 30% predicted)	Scoliosis surgery	Lung function (FVC) was assessed prior to planned surgery	Hospital discharge	Post operative complications including prolonged length of stay	No outcome difference was found between those patients whose FVC was above or below 30% predicted		Two patients (one in each group) needed tracheostomy. Length of stay and complication rate was similar for both groups. NIV was used in the immediate post-op period

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Martigne L, D Seguy, N Pelligrini, D Orlikowski	Efficacy and tolerance of gastrostomy feeding in Duchenne muscular dystrophy	Qualitative	2010 Clinical Nutrition	3	25	Duchenne Muscular Dystrophy boys aged 11-38 years (mean : 23yrs) gastrostomy between 1997 and 2007 NIV in 20 (14 to 27 yrs) 18 with tracheostomy (17.5 to 34.5 yrs)	Retrospective multicenter study of patients receiving gastrostomy within last 10 years with 2 months available weight measurements and followup	none	defined as from gastrostomy placement until death, removal of tube or end of clinical follow up (2 to 144 months, median 22 months)	body weight for age ratio based on centile chart for DMD boys (1988)before and after gastrostomy placement. complications associated with gastrostomy (<1 mth, > 1mth)	Patients(n=19) considered undernourished fell (n=11) ($p<0.05$) Wgt/Age ratio increased until 9 months post gastrostomy then plateaued (mean 69%pre - mean87%post) at max F/U 22mths($p<0.001$) 9/25 reached or exceeded mean wgt/Age ratio. Complications in 21 patients (84%)		gastrostomy can improve weight for age ratio though nutritional recovery incomplete or slow. Complication rate high and therefore debatable from this study whether truly reflects an improved quality or length of life.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow -up	Outcomes	Effect size	Funding	Comments
Martin, A. J. Stern, L. Yeates, J. Lepp, D. Little, J.	Respiratory muscle training in Duchenne muscular dystrophy	RCT	1986 Dev Med Child Neurol	1-	18	Patients with DMD with a mean age of 14.2 (range 7-20) years old. 94% were wheelchair bound	Patients were randomised to either group A or B. There was no significant difference between the two groups at baseline. Patients who were assigned to group A received IMT for 2 months followed by a 2 months of rest and then observed for 2 months. Those patients assigned to group B underwent no training for the first 2 months, rested for 2 months and then trained for the next 2 months. Training consisted of strength training (20% of Pi_{max} and Pe_{max}) and endurance (breathing through a mouthpiece with a resistance set that produced exhaustion within 3 minutes).	Changes in Pi_{max}, Pe_{max} or VC with training produced	6 months	No improvements with training	From beginning to end of the training period patients were able to tolerate a higher resistance for the 3 minutes ($p<0.01$).	unknown	A explanation for the lack of change in Pi_{max} and Pe_{max} is that the set level of 20% is too low to benefit the patient group.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Mellies, U, Ragette R, Dohna Schwake C, Boehm H, Voit T, Teschler H	Long-term noninvasive ventilation in children and adolescents with neuromuscular disorders	Qualitative	2003 European Respiratory Journal	3	30	Children (mean age 12.3 years) with various causes of neuromuscular weakness	NIV – started for daytime ventilatory insufficiency (n=14) or symptomatic SDB (n=16)	lung and respiratory muscle function tests, daytime arterial blood gas PSG with capnometry during nocturnal NIV	Upto 5 years	Improvement in nocturnal hypoventilation and daytime hypercapnia	At follow-up, 26 out of the 30 children reported using NIV for 8-12 hours per night. NIV improved or normalised nocturnal oxygen saturation and transcutaneous carbon dioxide levels (before 53.7+/-9.9 mmHg to 41.6+/-4.8 mmHg p<0.001). Daytime carbon dioxide levels were also normalised (PaCO2 before 47.5 +/-11.9 after 40.6+/-3.6 p<0.001). NIV use reduced respiratory disturbance (RDI before 10.5+/-13.1, after 3.1+/-3.5 p<0.001). 12 children made an attempt to stop NIV, but resumed on the first night because of symptoms. 10 children completed 3 nights without NIV. In all 10 children there was a prompt return of nocturnal hypoventilation with hypercapnia. All parameters returned to normal after 2 nights back on NIV.	Not stated	Provides evidence that NIV is effective in correcting sleep disordered breathing and daytime hypercapnia

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Mellies, U., R. Ragette, C. Schwake, H. Boehm, T. Voit and H. Teschler	Daytime predictors of sleep disordered breathing in children and adolescents with neuromuscular disorders	Prospective case series	2003 Neuromuscul Disord	2-	49 subjects of mean age 11.3 years	18 with congenital MD, 7 with Duchenne MD, 12 with SMA, 12 with other neuromuscular diseases	Lung function, blood gases, symptom questionnaire and polysomnography.	Between abnormalities of sleep breathing and the various daytime parameters	Not applicable	IVC < 40% predicts nocturnal hypercapnic hypoventilation. IVC < 60% predicts sleep breathing problems. PIP < 2.5 predicts hypercapnic hypoventilation. PaCO2 of more than 40 mmHg predicts sleep disordered breathing in children with a heterogenous group of muscle diseases.	Not applicable	University research grant and respiratory products company	Cross sectional study with a very mixed group of disease entities. This work suggests possible cut off points for further or more detailed sleep investigation.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow -up	Outcomes	Effect size	Funding	Comments
Mellies U, Dohna-Schwake C, Stehling F, Voit T.	Sleep disordered breathing in spinal muscular atrophy.	Qualitative	2004 Neuromuscul Disord.	3	12	All were children with SMA, 7 with and 5 without sleep disordered breathing (SDB) on PSG. All had VC <60%. The SDB was mild, and only one patient was hypercapnic.	NIV	The children with SDB were treated with NIV, the 5 children without SDB acted as controls	12 months	Repeat PSG and symptom score	Children in SDB group (RDI 14.1) were more likely to have symptoms (score 29 vs 15 $p<0.005$). These were improved after 6 months NIV – to the same level as found in the control group. RDI was also improved and same as control group (RDI 2.7 vs 2.2). Also noted improvements in weight (increase by 18% vs 5% in control group)	University of Essen, VitalAire Deutschland GmbH and Heinen and Lowenstein GmbH Alfried Krupp von Bohlen and Halbach Foundation.	Suggests that children with neuromuscular weakness and relatively minor sleep disordered breathing can get symptomatic improvement from NIV.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Meyer, T J, Pressman M R, Benditt J, McCool F D, Millman R P, Natarajan R, Hill N S.	Air leaking through the mouth during nocturnal nasal ventilation: effect on sleep quality	Qualitative	1997 Sleep	3	6	Adult patients established on NIV for nocturnal hypoventilation, one post TB, one with scoliosis, 4 with muscular dystrophy	Observational study of effect of mouth leak during nasal NIV	Sleep quality related to evidence of airleak	One nocturnal study and one daytime nap	Airleak – identified by high inspiratory flow through machine and confirmed with audio and videotape of the patients; correlated with PSG findings	Air leaks were found in all patients. 70-70% of arousals occurred after a period of leak, and the leak stopped temporarily after the arousal. Overall there was poor sleep quality with decreased amounts of slow wave and REM sleep. Oxygen sats were maintained by leak compensation in all but one patient.	Not stated	Doesn't prove the arousals were caused by the leak, but does suggest that the association is not due to fall in sats (as suggested by Bach when using volume targeted vents which do not compensate and so leak is associated with fall in sats.). Despite leaks being associated with sleep fragmentation, sleep quality is improved by NIV compared to no support.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow -up	Outcomes	Effect size	Funding	Comments
Miller G, O'Connor J	Spinal bracing and respiratory function in Duchenne muscular dystrophy	Qualitative	1985 Clinical Paediatrics	3	8	Letter. Children with DMD who were non-ambulant had lung function measured in and out of spinal braces	Bracing	Vital capacity	None	Vital capacity measured in and out of brace	Bracing reduced VC by a mean of 24% (4-37%)	Not stated	Small study.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow -up	Outcomes	Effect size	Funding	Comments
Miller, R. G., A. C. Chalmers, H. Dao, A. Filler-Katz, D. Holman and F. Bost	The effect of spinal fusion on respiratory function in Ducheene muscular dystrophy	Qualitative	1991 Neurology	3	39	39 patients from 1 DMD clinic, >9 yr old	17 underwent spinal surgery- segmental spinal fusion (Luque technique) 22 did not undergo surgery	Comparing the operated and un-operated gp in terms of lung function decline	Median follow up 34 months	Respiratory function Post op complications Sitting comfort Back pain	No sig differences between the 2 gps in terms of lung function decline in short term or up to 5 yrs follow up. Subjectively operated patients reported improvement in sitting comfort, appearance	Not stated	The statistical method for analysis of the data was not clearly stated in the paper. It appeared that all the lung function measurements for each gp were plotted for the regression against age. It would be more appropriate to examine the rate of change of lung function per patient in each group and then looking at the group change.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Miske LJ. Hickey EM. KolbS M Weiner DJ. Panitch HB.	Use of the Mechanical In-Exsufflator in Pediatric Patients with neuromuscular disease and impaired cough	Qualitative research	2004 Chest	3	62	62 patients (34male) median age 11.3 (range 3 months to 28.6 years) with the following diagnosis: DMD (n=17), SMA type I and II (n=21), Myopathy (n=12) and non specific NMD (n=12).29 patients recived tracheostomy ventilation and 25 used NIV	Retrospective review of the effect of patients who were provided with mechanical insufflation-exsufflation who had a PE_{max} less than 60cmH_2O	Respiratory muscle strength between diagnosis group. Tolerance of mechanical insufflation-exsufflation. Effect of mechanical insufflation-exsufflation on hospital admissions and chest xray	Patients had used mechanical insufflation-exsufflation for between 0.5-45.5 months	Mechanical insufflation-exsufflation was tolerated in 90% of patients. Chronic atelectasis resolved in 4 patients and 5 patients experienced a reduction in the frequency of pneumonias. One patient experience bradycardia.	Mean age (range) years for age of commencement of mechanical insufflation-exsufflation: DMD: 21.0 (16.7-27.7) Myopathy: 10.7 (0.2-22.9) Non specific NMD: 9.7 (0.3-18.5) SMA type I 2.4 (0.4-15.9) SMA type II 12.6(1.5-28.6) PE_{max} for each diagnosis mean±SD DMD: 21.1±3.0 Myopathy: 17.6±7.4 Non specific NMD: 10.4±2.4 SMA type II 15.5±5.5	Not reported	Mechanical insufflation-exsufflation was tolerated well in children with NMW. Children that benefited had a PEmax less than 30cmH2O

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Narayanaswami P, T. Bertorini, R. Pourmand, L. Horner	Long-term tracheostomy ventilation in Neuromuscular Diseases: Patient acceptance and Quality of life.	Qualitative	2000 Neurorehabilitation and Neural Repair.	3	19	DMD(8), ALS (8), other(3) aged 14-70 (mean: 40yrs) All receiving domiciliary tracheostomy IPPV. Mean duration 54 months.	Questionnaire on life satisfaction	Comparison of answers DMD : ALS patients		Demographics, duration/type of ventilator support, life satisfaction, effect of TIPPV on caregivers/families	13/19 were satisfied or very satisfied with life, 16 (84%) thought they had made the right choice in long term TIPPV. Satisfied patients were significantly younger than dissatisfied. DMD mean score 7: vs ALS mean score 3.25($p<0.01$)	Not reported	Small but useful study that addresses life satisfaction. Stresses the importance of informed decision making and implications for all family members including financial ones.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Niranjan V, Bach JR.	Non-invasive management of pediatric neuromuscular ventilatory failure	Qualitative	1998 Critical Care Medicine	3	10	Eight patient had DMD, 1 had SMA and 1 a cord lesion. Patients were ventilator dependent at admission, mostly with acute respiratory infection, although 2 were following surgery and one following trauma	Weaning from invasive ventilation by protocolised use of NIV	None	3.3 $\pm$ 0.9 years from ICU discharge		All 10 were successfully weaned from ventilation	Not stated	One of a number of studies from Bach's group looking at weaning strategies in NMW patients.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow -up	Outcomes	Effect size	Funding	Comments
Noble-Jamieson C, Heckmatt JZ, Dubowitz V, Silverman M	Effects of posture and spinal bracing on respiratory function in neuromuscular disease	Qualitative	1986 Archives of Disease in Childhood	3	40	All patients had neuromuscular disease. 20 were non-ambulant with scoliosis. Of the latter group 16 were wearing a brace to control scoliosis	None	Lung function (PEFR, FVC, FEV1) was compared between groups	None	Lung function measured sitting and supine	Non ambulant children had lower lung function and a greater reduction in supine vs sitting positions. Patients in a brace had a 22% reduction in FVC when wearing the brace.	Not stated	This study reported that in non-ambulant patients, lying down impairs lung function (by about 12%) but that a brace reduces lung function by a further 16%. Small numbers.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Nygren-Bonnier, M. Markstrom, A. Lindholm P. Mattsson E. Klefbeck B.	Glossopharyngeal pistoning for lung insufflation in children with spinal muscular atrophy type II	Qualitative research	2009 Acta Paediatrica	3	11	Children with SMA type 2, diagnosed at National Respiratory Centre, Danderyd. Sweden aged 6-16. years old. None had tracheostomies and all were free of any RTI	Children were taught glossopharyngeal breathing	1) Pre and post lung function 2) ability to learn glossopharyngeal breathing	8 weeks	IVC PEFR chest expansion at the level of the xiphoid process	45% (5 out of 11) of patients could learn GI Four of the children who completed the study showed a mean increase in IVC of 0.13 L (95% confidence interval (CI) 0.03-0.23) and PEFR of 116 L/min (95% CI 60-173). Chest expansion increased with glossopharyngeal breathing at the level of the xiphoid process of 1.50 cm (95% CI 0.16-2.84) and at the level of the fourth costa of 1.79 cm (95% CI 0.85-2.73). tension in the chest.	Health Care Sciences Postgraduate School.	Side effect of the technique: temporary symptoms of dizziness and. Patients who could learn the technique had improvements in IVC and PEFR

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Orlikowski D, Mroue G, Prigent H, Moulin C, Bohic M, Ruquet M, Raphael JC, Annane D, Lofaso F	Automatic air-leak compensation in neuromuscular patients: a feasibility study.	Qualitative	2009 Respiratory Medicine	3	14 adult patients each of whom had both modes of ventilation	neuromuscular disease (not specified) needing home ventilation (one with a trachy) who had symptoms of air leak	use of leak compensation (pressure targetted volume assured mode) using the VS Ultra ventilator (SAIME, Savigny le Temple, France	a night using leak compensation vs a night without leak compensation (random order)	Study was carried out over 2 nights	morning blood gases, patient tolerability (visual analogue score), sleep parameters	Improved morning CO2 with leak compensation (6.18 +/-0.9 vs 5.21+/-1.0 p=0.004) and improved tolerability. 9/14 patients stated they would like to continue with leak compensation at home. No difference in sleep parameters	French ministry of health	Most patients seem to cope well with simpler cheaper pressure targeted ventilators. Leak compensation modes may need to be reserved for patients with unresolved hypoventilation caused by leak. There was no EPAP used and this plus the need to measure expiratory TV means there needs to be an expiratory circuit and pneumotach. Risk of over-compensation if there is an expiratory leak - system was set to prevent dangerous overinflation so that Ti could only increase by 33% and pressure could only increase by +5cm

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Oskoui M, Levy G, Garland CJ, Gray JM, O'Hagen J, De Vivo DC, Kaufman n P.	The changing natural history of spinal muscular atrophy type 1.	Qualitative	2007 Neurology	3	143	Review of data held in the International Spinal Muscular Atrophy Patient Registry (90% of registrants are from the US). 391 patients sent questionnaires, 50% were returned.	Questionnaire on use of home ventilation, gastrostomy and cough assist.	Year of birth, use of NIV>16 hours, MIE and gastrostomy on survival	Retros pective review	Survival was main outcome	In a Cox proportional hazards model ventilation for more than 16 h/d (HR 0.3 (0.1–0.6) p=0.002), use of mechanical cough-assist device (HR 0.2 (0.1–0.5) p=0.001), and gastrostomy tube feeding (HR 0.5 (0.3–1.0) p<0.04) showed a significant effect in reducing the risk of death	Not stated	Provides evidence of improved survival in SMA1 – related to ventilation, MI-E, and gastrostomy feeding. Study has major drawbacks – requires families to register on voluntary database, and outcomes are not validated using hospital records or face to face interviews. Questionnaire required re-call of information in some cases from several years earlier. parents voluntarily signing up for a national registry and responding to questionnaires may be more likely to pursue proactive medical management – which may explain better survival in the 1980-1994 cohort as compared to other reports (eg Bach and Ioos)

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow -up	Outcomes	Effect size	Funding	Comments
Padman R, Lawless S, von Nessen S	Use of BiPAP by nasal mask in the treatment of respiratory insufficiency in pediatric patients: preliminary investigation	Qualitative	1994 Ped Pul	3	15	11 patients with neuromuscular diseases (2 SMA and 7 DMD) Aged 4 -21 yrs All present with respiratory failure on a background of chronic respiratory insufficiency without hypoxic	Use of NIV in acute respiratory deterioration for children with NMW and chronic respiratory failure	Physiological and biochemical status before and after NIV Hospital stay in the year prior to and after initiation of NIV	1 day - 21 months	Heart rate, respiratory rate , blood bicarbonate level, hospitalisation	Resting HR decreased ($p<0.01$) RR decreased ($p<0.01$) PaCO2 reduced ($p<0.02$) Hospitalisation reduced ($p<0.03$) Parents reported patients more comfortable at rest (80%); increased activity tolerance and ability to attend school (57%)	Not stated	The authors reported that NIV was a safe modality for treatment of respiratory failure albeit small number of patients This is a descriptive study (dated back to 1990-2) offering early paediatric data on NIV

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Pane M, Vasta I, Messina S,	Feeding problems and weight gain in Duchenne muscular dystrophy	Qualitative research	2006 European J of Paed neurol		118	Duchenne muscular dystrophy patients aged 13.8-35.8 yrs 32/118 ambulant 4/118 weight>2SD 5/118 . 10/118 (8%) gastrostomy placement at mean age 23.6 yrs (20-29 yrs) ,	Administration of questionnaires comprising a range of GI questions on feeding difficulties	none	N/A	Questionnaire on types of feeding difficulties, gastrointestinal symptoms, steroids and weight gain	12/118 (12%) had difficulties with mouth opening, 24/118 reported chewing difficulties and 21 reported choking less than once a week on average. Gastrostomy occurred in 10/118 (8%) at a mean age of 23.6 years.	None declared	Although choking was one of the most feared, it occurred infrequently even after the age of 18. Clinical signs of reflux required treatment in only 5. Chewing difficulties became increasingly frequent and mealtimes more prolonged. Aspiration pneumonia occurred in 7/118

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Parker D, Maddocks I, Stern LM	The role of palliative care in advanced muscular dystrophy and spinal muscular atrophy	Qualitative	1999	3	13	9 bereaved families, 4 current families of patients with NMD. Age at death of 9 patients ranged 8-31 yrs.	Semi structured interviews addressing experiences of support and discussion of End of Life care	N/A	N/A	Description of responses around common topics – Ventilator support, coordination of care, parent and sibling support, advance care directives and bereavement care	Concerns about paid carer issues. Timing and method of ventilator support discussions. Family and sibling support. 1/13 had written an advance care directive. 2/13 had received palliative care services	Crippled Children's Association Research and Development grant	Little knowledge around palliative care services with families. Timing and methods of discussion around ventilator support considered very important.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Petrone A, Pavone M, Testa MB, Petreschi F, Bertini E, Cutrera R.	Noninvasive ventilation in children with spinal muscular atrophy types 1 and 2	Qualitative	2007 Am J Phys Med Rehabil	3	9	All had SMA 1 or SMA 2 with asynchronous breathing pattern. (7 mos of age, range 2–33)	Use of NIV	Effect of NIV on chest and abdominal asynchrony	one study before NIV and one study 10 days later. In 5 patients 3 studies were done - one study before, one study on low-span BiPAP and one study on Hi-span BiPAP	phase angle plus sleep parameters	phase angle improves from 127 (73-152) to 44 (38-68)(normal). mean CO2, AHI, number of desats also improve. Improvement in phase angle only seen if Hi-span used - in the 5 patients who had span phase angle improved to 88 (54-110) with same improvement in all the other parameters; using hi-span, no further benefit in CO2 or Sats, but phase angle better - 48 (38-68)	Not stated	In young children with SMA 1 and SMA 2 with asynchonous chest and abdominal wall movement caused by intercosal muscle weakness, night-time abnormalities on sleep study can be corrected with low-span BiPAP, but full correction of the asynchony is only achieved with hi-span bipap. This may be important to prevent subsequent chest wall deformity.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Phillips, M. F., P. E. Smith, N. Carroll, R. H. Edwards and P. M. Calverley	Nocturnal oxygenation and prognosis in Duchenne muscular dystrophy	Retrospective cohort	1999 Am J Resp Crit Care Med	3	18 patients with DMD, older than 14 years at study entry	All DMD	Baseline spirometry, blood gas tensions, maximal respiratory pressures and BMI measured	Between the various measures and length of survival from study entry.	Followed over 10 years	Age of loss of ambulation and BMI do not predict duration of survival or age at death. PaO2 minimum nocturnal value correlates with survival in adolescents with DMD. PaCO2 correlates with VC. PaCO2 correlates with survival. Minimal nocturnal SaO2 correlates with survival	R values between 0.65 and 0.72	Partly by UK Muscular Dystrophy Group	Provides evidence that blood gas estimation can provide a guide to prognosis.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Phillips, M. F., R. C. Quinlivan, R. H. Edwards and P. M. Calverley	Changes in spirometry over time as a prognostic marker in patients with Duchenne muscular dystrophy	Retrospective study of subjects with at least 2 years of follow-up.	2001 Am J Resp Crit Care Med	3	58 patients with DMD	All DMD, at least 10 years old at study entry.	Describes rate of PFT decline	Between repeated spirometry values and survival	Variable follow up length. Mean age at death 21.5 years	Age when VC falls below 1 litre is strong marker of mortality. Maximal VC recorded and its rate of decline predicts survival.	Not applicable	Muscular Dystrophy Campaign (UK)	One of the few good studies with repeated measurements allowing more detail of rate of decline and change in lung function.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Philpot J, Bagnall A, King C, Dubowitz V, Muntoni F	Feeding problems in merosin deficient congenital muscular dystrophy	Qualitative	1999 Arch Dis Child	3	14	Children aged 2-14 years with merosin deficient congenital muscular dystrophy	Case series of prevalence and type of feeding difficulties with video-fluoroscopy and pH studies on symptomatic patients	N/A	6 years	Reported feeding difficulties, weight measurement, SALT assessment, video-fluoroscopy and pH studies	12/14 <3rd centile all reported chewing difficulties, 13 prolonged mealtimes 13/14 abnormal video-fluorscopy, 6/8 GO reflux, 5 gastrostomy which improved weight gain and stopped chest infection	Not reported	High prevalence of feeding difficulties in this small population of CMD. Descriptive study only.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Piasecki JO, Mahinpour S, Levine DB	Long-term follow-up of spinal fusion in spinal muscular atrophy	Qualitative	1986 Clinical Orthopaedics and related research	3	17	Children with SMA who had scoliosis surgery	Descriptive study of the outcome of surgery in this group	Degree of surgical correction and VC (nine patients studied 7 years following surgery)	Up to 7 years	Lung function (nine patients), clinical status	The study showed good qualitative outcomes with improved sitting and balance. Inadequate data was provided to evaluate the long term effect on lung function.	Not stated	A descriptive study with limited data. However there are few data on the outcome of surgery in SMA. The authors reported few respiratory complications of surgery.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow -up	Outcomes	Effect size	Funding	Comments
Piastra M, Antonelli M, Caresta E, Chiaretti E, polidori G, Conti G.	Noninvasive ventilation in childhood acute neuromuscular respiratory failure: a pilot study	Qualitative	2006 Clinical investigations	3	10	10 non-consecutive children with NMW in acute respiratory failure Heterogenous gp of NMD including mitochondrial myopathy and cerebellar ischaemia Aged 3m -12yr	The use of NIV in averting intubation for children in ARF	Blood gases before and after initiation of NIV	Hospital discharge	PaCO2 PaO2 FiO2	NIV successful in 8 of 10 children. In successfully treated pt: PaO2/FiO2 increased at 3 and 12 hr after NIV ($p<0.001$) Alveolar-to-arterial oxygenation difference also decreased ($p<0.001$) and hypercarbia normalised within 6-8 hours	Not stated	Authors concluded that NIV was a safe, effective first line therapeutic option for children with NMW and acute resp failure There was no description of criteria for initiating NIV (in terms of threshold for ABG parameters)

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow -up	Outcomes	Effect size	Funding	Comments
Pope JF, Birkrant DJ	Noninvasive ventilation to facilitate extubation in a pediatric intensive care unit	Qualitative	2000 J Intensive Care Med	3	25	Patients aged 0.2 – 19 years. Four patients had SMA I, 2 had SMA II and 4 had DMD	Using NIV to transition patients off mechanical ventilation	none	Not stated but at least the duration of hospital stay	Successful NIV assisted extubation Reintubation tracheostomy	NIV assisted extubation was successful in 7/10 patients with NMW on first attempt and two more at the second attempt. The remaining patient required tracheostomy Risk factors for failure of extubation included ineffective cough, failure to manage respiratory secretions, severe upper airway obstruction, impaired mental state	Not stated	Heterogenous group of patients including NMD and non NMD patients Success rate comparable to adult studies

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Ragette, R., U. Mellies, C. Schwake, T. Voit and H. Teschler	Patterns and predictors of sleep disordered breathing in primary myopathies	Prospective case series	2002 Thorax	2-	42 adult patients, mean age 28.7 years	10 with Duchenne MD, 10 with congenital MD, 7 with limb girdle dystrophy, 12 with acid maltase deficiency and 3 others with myopathies	Upright and supine IVC, maximal inspiratory muscle pressure, respiratory drive, respiratory muscle effort and arterial blood gases	Between daytime measures and polysomnography with capnography.	Not applicable	IVC < 25% predicts respiratory failure. IVC < 40% predicts nocturnal hypercapnic hypoventilation. IVC <60% predicts sleep disordered breathing. PIP < 4 predicts need for ventilation.	Not applicable	University research grant	Data is all in adults and in a very heterogeneous group. It does provide potential cut off points within daytime measures that are correlated with sleep breathing problems.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Rahbek J, Werge B, Madsen A, Marquardt J, Steffensen BF, Jeppesen J	Adult life with Duchenne muscular dystrophy : Observations among an emerging and unforeseen patient population	Qualitative	2005 Pediatric Rehabilitation	3	65	Danish adults with Duchenne muscular dystrophy aged 18-42 years	Semi-structured questionnaire exploring quality of life, social participatory and body functional profiles through home visit and interviews	N/A	N/A	Description with overall and age-specific distribution of responses	70.7% invasively ventilated, 18.5% NIV, 10.8% unassisted ventilation.52.3% live alone, 44.6% with parents. 38.5% pains daily restricting activity in 9 subjects. High school completed in 9 subjects. 52.4% reported fatigue, which restricted activity in 11 subjects. Sexual problems in 37 subjects. Excellent quality of life reported in 83.1% all subjects with 75.4% reporting “no or few worries” about the future.	Not reported	In spite of a good quality of life, lack of further education, occupational activity, sexual experience and restriction in social participation with prevalence of daily pains in nearly 40%.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Ramelli G.P, A. Aloysius, C King, T Davis and F Muntoni	Gastrostomy placement in paediatric patients with neuromuscular disorders : indications and outcome	Qualitative	2007 Dev Med and Child Neurol	3	31	15 cong Muscular dystrophy 11 cong myopathies 6 other NM diagnoses 17 males and 15 females age : 2y8mo to 31 yrs (mean 12yr8mo)	descriptive series of indications and outcome of gastrostomy in NM disease	none	N/A retrospective notes review	growth, height/weight before and after gastrostomy placement no. chest infections before and after	No stats. improvement in weight centiles and height centiles 1 yr post placement. 17/29 chest infections prior to procedure, 15 > 6 episodes per year) 26/29 no further chest infections at time of case note review.	nil reported	case series showing gastrostomy to be a useful intervention to reverse faltering growth in this population. and with associated reduced number of infections reducing the frequency of hospitalisation.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Raphael JC, Chevret S, Chastang C, Bouvet F.	Randomised trial of preventive nasal ventilation in Duchenne muscular dystrophy. French Multicentre Cooperative Group on Home Mechanical Ventilation Assistance in Duchenne de Boulogne Muscular Dystrophy	RCT	1994 Lancet	1-	70 (35 in each group) Planned to have 100 patients, 50 in each group, but trial stopped before full recruitment because of excess deaths in the treatment arm.	Boys with DMD of any age with FVC between 20 and 50% predicted but with daytime PaO2>60mmHg and daytime PaCO2<45; if the boys were over 15 and still able to walk, they were also excluded.	Randomised to NIV plus conventional treatment, or to conventional treatment alone. NIV was delivered at night for a minimum of 7 hours and at a tidal volume of at least 10ml/kg. Volume cycled controlled ventilation was used.	Follow-up was every 6 months. ABGs and lung function were measured.	5 years or death.	Death was primary outcome. Secondary outcome measures were: daytime hypercapnia Fall of FVC by 20% from baseline Meeting criteria for needing NIV - these are not clearly stated, but probably FVC<20% OR daytime PaO2>60mm Hg OR daytime PaCO2<45	There were 8 deaths in the treatment group (of which 5 were clearly respiratory failure) and 2 deaths in the control group (of which one was clearly respiratory failure).	Institut National de la Sante et de la Recherche Medicale and Association Francaise de lutte contre les Myopathies	No benefit in prophylactic NIV, and suggestion that false sense of security from having NIV at home increased risk of death. There are some limitations to the study: we don't know if the patients had symptoms of nocturnal hypoventilation; there were no sleep studies done, and no way of assessing the effectiveness of the NIV; compliance with ventilation was assessed only by questionnaire. No improvement in time to daytime hypercapnia suggests that the NIV was not as effective as it might have been in correcting daytime hypercapnia.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Read J, Simonds A, Kinali M, Muntoni F, Elena Garralda M.	Sleep and well-being in young men with neuromuscular disorders receiving non-invasive ventilation and their carers	qualitative	2010 Neuromuscular Disorders 20:458-463	3	10	Males with neuromuscular disorders (aged 12-25 years) receiving nocturnal ventilation at a specialist centre. Mean age starting ventilation : 15,4+/-3.8 yrs)	Questionnaires to patients (n=10) and carers (n=9) and interviews – scores compared to values for general population	N/A	N/A	Pittsburgh Sleep quality index, Short form 36 (SF-36) measure of wellbeing Hospital anxiety and depression scale (HADS) Family Burden Interview Schedule, Family assessment device	Pittsburgh scale (sleep quality) was 7.5/21 (+/- 4.2), 90% above cutoff for poor sleep. But mean sleep satisfaction scores high (7.2+/-2.7) SF-36 for social functioning 62.5 vs 91.5 (general pop) Mental health 70.8vs 79.6 (gen pop), vitality 58.5vs 86.0 (gen pop). No excess of anxiety or depression on HADS. Mean carer sleep index scores : 7.4+/-4.7 (70% above cut off for poor sleep). No excess of anxiety on HADS but more than expected had higher depressive scores (11% vs 3% gen pop). 1/3 families reported moderate/high levels of burden on family life, health and finances	Muscular Dystrophy campaign Centre grant to Dubowitz Neuromuscluar centre.	Qualitative and quantitative data on both patient and carer about sleep quality, general wellbeing and mental health Comparing results to general population. Sleep quality was porr causing particular issues for carers, linked to significantly poorer emotional wellbeing and greater family burden.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow -up	Outcomes	Effect size	Funding	Comments
Reardon CC, Christiansen D. Barnett ED Cabral HJ.	Intrapulmonary percussive ventilation vs. incentive spirometry for children with neuromuscular disease	RCT	2005 Arch Pediatr Med.	1-	18	Median age for each group was 17 years, with age ranges for IPV (11- 19) years and for IS (14- 19) years.. One subject in each group received nocturnal non-invasive ventilation for chronic respiratory failure related to underlying NMD	IPV or incentive spirometry	IPV versus IS PFT pre and post and CI	7 months	PFT showed no change days off school antibiotic use supplemental treatment Spiromertry and lung volumes. Use of antibiotics and missed days of education.	Patients in the IPV group did not receive any antibiotics during the study period. Patients in the IS group received 44 days of antibiotics.). Patients in the IS group (n = 3) had 3 episodes of pneumonia or bacterial bronchitis compared with none for patients in the IPV group (IRR, 3.9; 95% CI, 0.43-35). The IS group required significantly more supplemental respiratory treatments than the IPV group:	Deborah Munroe Noonan Memorial Fund, Boston, Mass; intrapulmonary percussive ventilation devices provided by Percussionaire Corp, Sandpoint, Idaho; Parker B. Francis Fellowship Award, Harvard School of Public Health, Boston (Dr Reardon).	Intrapulmonary percussive ventilation as part of a preventative pulmonary regimen reduced days of antibiotic use and hospitalisation for respiratory illness in adolescents with neuromuscular disease.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow -up	Outcomes	Effect size	Funding	Comments
Robins-Miller J, A Colbert, J Osberg.	Ventilator dependency ; decision making, daily functioning and quality of life for patients with muscular dystrophy	qualitative	1990 Dev Med and Child Neurol	3	17	17 patients with Duchenne MD aged 17-33 yrs average ventilator use 5yrs (1-11 yrs) 14 family members – mothers aged 32-64	Decision making about ventilation, daily functioning before and after ventilation, educational activity – before and after		N/A	Recall pre and post ventilation on decision making, daily functioning and educational activity	16/17 had <6 mths to make decision re : ventilation 11/17 patients and 6/14 families felt quality of life had declined. Educational activity had been negatively affected	Research grant – National Institute of Disability and Rehab Research, Washington	Timely education required about progression and potential impact upon daily functioning in order to make objective decision.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow -up	Outcomes	Effect size	Funding	Comments
Robinson D, Galasko CS, Delaney C, Williams on JB, Barrie JL	Scoliosis and lung function in spinal muscular atrophy	Qualitative	1995 European Spine Journal	3	43	Patients with SMA. 16 went on to have scoliosis surgery	None	Rate of progress of scoliosis; lung function following surgery	1 year	FVC, Cobb angle	FVC had an inverse relationship to the Cobb angle. FVC was improved following surgery.	Not stated	Retrospective study and the only study to report improvement in FVC following surgery in NMD

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Rodillo, E. Noble-Jamieson, C. M. Aber, V. Heckmatt, J. Z. Muntoni, F. Dubowitz, V.	Respiratory muscle training in Duchenne muscular dystrophy	RCT	1989 Arch Dis Child	1-	22	9-14 years olds with DMD	Double blind crossover trial of IMT versus PEFR	IMT with an incentive spirometer (20 inspiratory manoeuvres a day) or placebo training with a peak flow meter (10 peak flows).	18 days	FEV1, FVC, PEFR and Pi_{max}	PEFR improved during placebo training ($p<0.02$) and Pi_{max} during IMT ($p<0.02$).	Muscular Dystrophy Group	No benefit was seen in training with spirometry or PEFR manoeuvres

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow -up	Outcomes	Effect size	Funding	Comments
Rotimi F.,Childs A.-M.,Potrata B.	A pilot study to evaluate current practices of transition of young adults from paediatric to adult neuromuscular (NM) clinics	Qualitative	2010 European J Child Neurol. 17/(532), 1351-5101 (poster)	3	21	21/71 young people aged 16-25yrs with NM disorder and 17 carers from Leeds population attending a tertiary NM clinic	Evaluation of current practice of transition, identification of key needs By questionnaire and 2 focus groups	Review against Dept of Health guidelines on transition	N/A	Descriptors only of key themes	Inadequate information, Age preference -18 years Longer duration at transition clinic,specific adult NM clinic, identification of a key worker, current transition practice was too abrupt	Not reported	

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow -up	Outcomes	Effect size	Funding	Comments
Seeger BR, Sutherland AD, Clark MS	Orthotic management of scoliosis in Duchenne muscular dystrophy	Qualitative	1984 Arch Phys Med Rehabil	3	24	Boys with DMD age from 6 to 20 years	Spinal support. 4 groups: unmodified wheelchair, modular seating, spinal jacket and custom-molded seat. Jackets were wron 7 hours per day.	Progression of scoliosis	Mean of 3 years	Rate of progression of scoliosis	6 monthly xrays were taken to monitor the rate of scoliosis progression. There was no difference in the rate for those patients treated with a spinal jacket or modified seat compared to an unmodified wheelchair.	Not stated	A descriptive paper of practice in a single unit on a very small number of subjects. There are no numbers given for each of the subgroups, nor any reason why different treatment modalities were chosen.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Sakakihara Y, M. Kubota, S. Kim, A Oka	Long-term ventilator support in patients with Werdnig-Hoffman diesease	qualitative	2000 Pediatrics International	3	33	paediatricians from 31 hospitals across Japan looking after 36 ventilated patients with WHD aged 2 mths to 13 years (mean 5yrs 6 mo)	postal questionnaire		N/A	attitudes of Paediatricians in Japan towards ventilator assistance for type 1 SMA patients	mechanical ventilation as an emergency in 12/32 24/30 thought quality of life was inadequate 17/30 would start ventilator assistance if a new patients with WHD Parents opinion (n=15) greatest in decision making	not reported	strong fanily endorsement is a key factor in favouring life support treatment for patients with WHD in Japan. Confinement to hospital was most commonly cited reason for unsatisfactory quality of life.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow -up	Outcomes	Effect size	Funding	Comments
Seguy D, L Michaud, D Guimber, J-M Cuisset	Efficacy and tolerance of gastrostomy feeding in paediatric forms of neuromuscular disease.	Qualitative	2002 J of Parenteral and Enteral Nutrition	3	12	10 male and 2 female age range : 1 month to 25.5 years All clinical evidence NMD	Evaluation of nutritional effects/ tolerance of gastrostomy	none	6 months to 1 year or to end of gastrostomy feeding	Z scores weight for age and height both before and after. complications described	W/A and W/H Z scores improved for all 10 patients W/A $p<0.05$ and W/H $p<0.001$	not reported	gastrostomy feeding is well tolerated with a measured significant improvement in weight

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow -up	Outcomes	Effect size	Funding	Comments
Servera, E., J. Sancho, M. J. Zafra, A. Catala, P. Vergara and J. Marin	Alternatives to endotracheal intubation for patients with neuromuscular diseases	qualitative	2005 Am J Phys Med Rehab	3	17	11 ALS; 4 DMD; 1 transverse myelitis; 1 non-specific myopathy) 10M; 7F All patients are in acute respiratory distress agreeing to NIV instead of ETT	Continuous NIV and cough aids, to avert intubation	Comparisons were made between the successfully treated and unsuccessfully treated group in terms of risk factors including age and lung function	Up to 3m post hospitalisation	ABG at admission and end of stay Duration of continuous NIV Length of stay in hospital Need for intubation and tracheostomy death	19/24 episodes (79.2%) averted intubation No sig differences in lung function between the successfully treated and unsuccessful group Bulbar dysfunction was an independent risk factor for failure of NIV ($p<0.05$; odd ratio 35.99%; 95% CI 1.71-757.68)	Not stated	The study concluded that intubation can be avoided for some patients with NMD in acute resp failure by NIV and cough assist Prospective study BUT this is an adult study

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow -up	Outcomes	Effect size	Funding	Comments
Shapiro F, Sethna N, Colan S, Wohl ME, Specht L	Spinal fusion in Duchenne muscular dystrophy: a multidisciplinary approach	Qualitative	1992 Muscle & Nerve	3	27	27 children with DMD	Spinal fusion	Pre and post op lung function	Mean follow up post surgery 40 months	Lung function pre and post op Post op complications	The mean forced vital capacity (FVC) preoperatively was 45.3 +/- 15.9% with continuing diminution to 28.7 +/- 14.9% at 3.3 +/- 2.2 years after surgery.	Not stated	This study did not show any significant long term increase or stabilisation of lung function after spinal fusion The authors also described the results of multidisciplinary pre-op assessments including orthopaedic, anaesthetic, cardiology and respiratory assessments.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow -up	Outcomes	Effect size	Funding	Comments
Shinonaga C, Fukuda M, Y Suzuki, T Higaki et al	Evaluation of swallowing function in Duchenne muscular dystrophy	Qualitative	2008 Dev Med and Child Neurol	3	5	DMD patients aged 18-24 years 3 needing NIPPV during sleep	Case series of reported feeding difficulties, assessment and video-fluoroscopy	N/A	N/A	Descriptive only from objective testing and reports	Not measured quantitatively. Delayed pharyngeal transit time but size of effect not measured	Not reported	Feeding dysfunction is common and under reported

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow -up	Outcomes	Effect size	Funding	Comments
Simonds AK, Muntoni F, Heather S, Fielding S.	Impact of nasal ventilation on survival in hypercapnic Duchenne muscular dystrophy	Qualitative	1998 Thorax	3	23	Consecutive patients with Duchenne muscular dystrophy at a single centre, started on NIV either following acute respiratory failure (n=5) or symptomatic daytime hypercapnia AND nocturnal hypoventilation.	Nocturnal NIV	None – observational study	Up to 5 years	Survival, quality of life (using SF-36), blood gases.	All patients tolerated NIV and none asked to discontinue it. Survival 73% at 5 years. Blood significantly improved. Quality of life ratings not directly affected by mobility issues did not differ from age and sex matched controls.	Not stated	There was no comparison group in this study, but survival is favourable compared to historical controls including the untreated group in the Vianello et al study.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow -up	Outcomes	Effect size	Funding	Comments
Simonds AK, Ward S, Heather S, Bush A, Muntoni F.	Outcome of paediatric domiciliary mask ventilation in neuromuscular and skeletal disease.	Qualitative	2000 Eur Respir J	3	40	Children with ventilatory insufficiency due to neuromuscular weakness aged 9 month to16 yrs. Started on NIV for symptomatic nocturnal (18); hypoventilation, diurnal ventilatory failure, (17); following acute respiratory failure (3); frequent chest infections associated with sleep-disordered breathing (2)	NIV	None – observational study	Mean of 30 months	Daytime and night-time blood gases.	Daytime Pa,O2 increased from 8.5+/- 1.8 to 10.9+/-1.7 kPa ($p<0.001$) and mean Pa,CO2 from 7.0+/-1.6 to 5.9+/-0.8 kPa ($p=0.01$). Nocturnal mean SaO2, minimum SaO2 and maximum tPCO2 improved from 65+/-19.2%, 85.2+/-7.2% and 8.8+-2.15 kPa to 84+/-8.6%, 92.4+/-2.7% and 7.6+/-1.4 kPa ($p=0.03$).	Not stated	Uncontrolled study showing that NIV can correct nocturnal hypoventilation and daytime hypercapnia. No comment on whether symptoms resolved, but assumption is that they did.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Socrates C., Grantham-McGregor S.M., Harknett S.G., Seal A.J	Poor nutrition is a serious problem in children with cerebral palsy in Palawan, the Philipines	Qualitative cross sectional study	2000 Int J Rehabil Res	3	115 children	31 with cerebral palsy, 20 siblings and 64 neighbourhood children	Measurements of height, weight and armspan	Between disabled children and both groups of controls	Not applicable	Height was estimated in the disabled group by using predictive equation derived from measurements in the controls.	Not applicable	Not stated	Equation generated that allowed height to be estimated from armspan measurement.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Soudon P, Steens M, Toussaint M.	A comparison of invasive versus noninvasive full-time mechanical ventilation in Duchenne muscular dystrophy.	Qualitative	2008 Chron Respir Dis	3	42	All had DMD, needing respiratory support for a minimum of 15 hours per day. Retrospective study looking at complications. 16 had trach, 26 had NIV. Not randomised and the trachy patients were 5 years older and had lower VC. 8/16 trach and 10 out 26 NIV patients died in 5 year period. Both groups had been using diurnal ventilation for 3 years.	None – retrospective study	Tracheostomy ventilation versus mask ventilation	None – retrospective data collected only	Complication rates	Equal mortality (despite older age of trach patients). Trach patients had complications directly related to trach – 60% had granulomas (trach had been in place for mean of 3 years at start of study period) – requiring either steroids or laser surgery, had more secretions, had more recurrent chest infection and were more likely to need care in a rehab hospital as compared to home or a nursing home. Patients with NIV were more likely to have low weight and need g-tube feeding. This reflects requirement to stop mouth-ventilation when trying to swallow.	Not stated	Suggests that where possible NIV should be used for diurnal ventilation when daytime resp failure occurs despite nocturnal ventilation. There is not an inevitable progression from NIV to trach. If mouth ventilation is used, need to consider earlier g-tube to prevent weight loss.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Suresh S, Wales P, Dakin C, Harris M, Cooper D	Sleep related breathing disorder in Duchenne muscular dystrophy : Disease spectrum in the paediatric population	Qualitative	2005 J Paediatr. Child Health	3	34	Duchenne MD patients aged 1-15 years referred for respiratory assessment FVC between 12 and 107% predicted. (n=29). M=33, F=1	Retrospective notes review Polysomnography conducted in 32 patients	N/A	Data available for 5 year period	Polysomnography findings – sleep efficiency, obstructive event index, AHI, lowest O2 sat, %sleep time <90%O2sat, highest tCO2, FVC	22(64%) reported sleep related symptoms 10(31%) diagnostic of OSA(median age 8 years) 11(32%) hypoventilation (median age 13 yrs) with median FVC of this group 27% predicted. All had adenotonsillectomy – repeat polysomnography in 6/10 showed improvement but not statisically significant Improvement in AHI with NIV (mean difference =11.31,95%CI 5.91-16.70, P=0.001.). No association between symptoms and sleep disordered breathing.	None declared	Significant OSA in this population – successfully treated with adenotonsillectomy though small numbers meant not statistically significant. Occurs generally in 1st decade (median age 8 years)

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Sy K, S Mahant, N Taback, J Vajsar, P G Chait, N Friedman	Enterostomy tube placement in children with spinal muscular atrophy type 1	Qualitative	2006 J of Paediatrics	3	12	all SMA type I hospital based tertiary centre	complications in first 30 days after enterostomy placement	none	30 days	pneumonia, respiratory failure, unplanned ICU admission rates	7/12 patients no major complications mortality rate 2/12 (16.7%) 5/12 aspiration pneumonia in older patients (41.6%) 4/12 required PICU admission.	not reported	suggests best done early if to be undertaken but needs careful evaluation of quality of life benefits alongside aims of such an intervention.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Takaso, M., T. Nakazawa, T. Imura, K. Fukushima, W. Saito, R. Shintani, G. Miyajima, M. Itoman, K. Takahashi, M. Yamazaki, S. Ohtori, M. Oka and A. Sasaki	Surgical management of severe scoliosis with high-risk pulmonary dysfunction in DMD	Qualitative	2010 International Orthopaedics	3	14	14 DMD (aged 11-17 yr) patients with severe scoliosis and poor FVC <30%	Scoliosis surgery Respiratory muscle training pre-op	n/a	2 yrs	Respiratory complications Post operative scoliosis Changes in FVC post op	No patient developed respiratory complications FVC increased v slightly or remained stable in all pt at 6 wk post op (not statistically sig) At 2 yrs post op = FVC continued to decline Maintenance of curvature correction at 2 yrs	Not stated	Small series Favourable results with pulmonary muscle trainer pre op

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow -up	Outcomes	Effect size	Funding	Comments
Tangsrud , S. E., K. C. Carlsen, I. Lund-Petersen and K. H. Carlsen	Lung function measurements in young children with spinal muscle atrophy; a cross sectional survey on the effect of position and bracing	Qualitative	2001 Arch Dis Child	3	8	8 children with SMA I and II	Bracing	Comparing tidal volume loop and passive respiratory mechanics in sitting and supine position , with and without bracing (in 5/8 children)	Not stated	Tidal expiratory volume Compliance of the respiratory system	Tidal expiratory volume and compliance of the respiratory system are higher in sitting than supine position (not sig) Bracing in sitting position reduced tidal expiratory volume	Not stated	Bracing may impair tidal respiration in children developing scoliosis in SMA Small case series; no controls

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow -up	Outcomes	Effect size	Funding	Comments
Thompson R.J, J Zeman, D Fanurik, M Sirotkin-Roses	The role of parent stress and coping and family functioning in parent and child adjustment to Duchenne muscular dystrophy	qualitative	1992 J of Clinical Psychology	3	35	Age 57-177 mths, 34 Caucasians, 1 hispanic, boys with Duchenne MD. Socioeconomic status	To investigate psychosocial factors associated with DMD in children through self-report measures and interview	N/A	N/A	Parental appraisal of stress. Ways of coping questionnaire Family Environment Scale. Symptom checklist 90-Revised. Missouri Children's behavior checklist	57% of parents self-reported poor psychosocial adjustment. 89% rate of parent – reported behavior problems in children. High parent use of palliative coping methods and high levels of family conflict were associated with poor adjustment.	Not reported	High self-reported stress amongst parents, poor psychosocial adjustment. High levels of parent-reported behaviour problems

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Tompsett J., Yousafzai A.K., Filteau S.M.,	The nutritional status of disabled children in Nigeria: a cross-sectional study	Cross sectional study	1999 Eur J Clin Nutr	3	311 children, all under 10 years of age	112 with various disabilities, 87 siblings and 112 neighbours	Measurements of height, weight, mid upper arm circumference, demispan and halfspan	Between those with disabilities and controls	Not applicable	Measurement difficulties described in disabled children. Halfspan was found to be a use proxy for height in these children	Not applicable	Grant from TEAR Fund	Paper describes height and weight for age as being reduced in the disabled children. Provides evidence in a different population that measures of armspan can act as a useful proxy to estimate height.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Topin, N. Matecki, S. Le Bris, S. Rivier, F. Echenne, B. Prefaut, C. Ramonatxo, M.	Dose-dependent effect of individualized respiratory muscle training in children with Duchenne muscular dystrophy	RCT	2002 Neuromuscul Disord	1-	18	Children with DMD	Endurance training (30% of their P_{imax})	Endurance training (30% of their P_{imax}) versus placebo8 DMD training (5% of their P_{imax}) for a	six week period	inspiratory muscle endurance (T_{lim})	Endurance training (307.6 ± 126.6 to 448.4 ± 176.7(46% increase)) ($p<0.05$) versus in the placebo group (271.1 ± 40.4 to 271.7 ± 45.6) (p=ns)	This investigation was supported by the ‘Association Franc¸aise contre les Myopathies’ (AFM), (grant no 5395).	Endurance training increase inspiratory muscle endurance in patients with DMD who are trained at 30% P_{imax}

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Toussaint M De Win H. Steens M. Soudon P.	Effect of intrapulmonary percussive ventilation on mucus clearance in Duchene muscular dystrophy patients: a preliminary report	Qualitative research	2003	2	8	DMD patients all with tracheostomies advanced stage with severe respiratory insufficiency VC< 600ml PEF<150L/mmin cardiovascular stable	The effect of addition of IPV on secretion clearance	Random cross over of 15 minute treatment regime conventional treatment (FET, MAC + et suction then aerosol therapy) vs. conventional treatment + IPV during aerosol therapy stage	Patients stayed in hospital for 5 days to complete the study	secretion weight SpO2 HR PEF PeTCO2 airway resistance	patients who were hyper secretive (n=5) produced more secretions during IPV T0 3.87+/-3.33 vs. T1 and 2 6.5 +/- 4.77 p=0.01 IPV - T0 4.27+/-3.04 vs. T1and 2 4.57 +/-3.5	In house	IPV is safe and in patient with secretions may be of benefit but further studies are warranted

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow -up	Outcomes	Effect size	Funding	Comments
Toussaint , M., M. Steens, G. Wasteels and P. Soudon	Diurnal ventilation via mouthpiece : survival in end-stage Duchenne patients	Qualitative research	2006 European Respiratory Journal	3	42	All had DMD, needing resp support at night, and then developing daytime hypercapnia. Study looking at resp scores and VC over 7 year period. 3 patients excluded – one with intellect impairment, 2 with recurrent aspiration – all 3 had trach.	Use of mouth-piece ventilation.	None	7 years	VC, Max insp pressure (MIP) PCO2 and resp scores.	VC fell, but apparently not significantly (500 to 350). MIP fell dramatically (16 to 6). Max time off any support fell from 15 hour to 30mins. Appetite, dyspnoea and swallowing improved in majority and symptom score fell from 3.2/7 at MIPPV onset to 0.8/7 1 year after MIPPV onset. Daytime CO2 at onset was 60mmHg. It was normalised by MIPPV and stayed normal. Assist control volume ventilation used with mean tidal volume of 688ml. Survival was 50% at 31 years.	Not stated	MIPPV is effective despite concerns about mouth leak because of weakness – patients can accommodate this and will purse their lips when then need a bigger breath. MIPPV reverses symptoms associated with daytime resp failure (dyspnoea; loss of appetite and weight; depression; intellectual fatigue or trouble of concentration; headaches; difficulty swallowing)

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Toussaint, M., M. Steens and P. Soudon	Lung function accurately predicts hypercapnia in patients with Duchenne muscular dystrophy	Cross sectional	2007 Chest	3	114 patients	Duchenne MD All in wheelchairs All over 12 years old	Lung function and day and night gas exchange were determined	Grouped into those with night-time hypercapnoea only, those with diurnal hypercapnoea and those who were eucapnic	Not applicable	VC < 680 mls correlates with daytime hypercapnoea. VC < 1820 mls correlates with nocturnal hypoventilation.	ROC 0.968	Not stated	Relationship between loss of daytime lung function as determined by VC and loss of ability to maintain normal blood gases.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Toussaint M. Boitano, LJ. Gathot, V Steens M. Soudon P.	Limits of effective cough-augmentation techniques in patients with neuromuscular disease	Qualitative research	2009 Respir Care	3	179 Maximum insufflation capacity and maximum insufflation capacity combined with a manual assisted cough was measured in a subgroup of 60 patients on NIV.	Patients >8 years with diagnosis: DMD n=117 Becker muscular dystrophy n=10 SMA type II n=26 Miscellaneous n=26 60% patients required NIV	Manual assisted cough, Maximum insufflation capacity, Combination of maximum insufflation capacity with a manual assisted cough	Changes in PCF from a baseline unassisted cough then in the following 3 situations: Manual assisted cough, Maximum insufflation capacity, Combination of maximum insufflation capacity with a manual assisted cough	One off visit	Changes in PCF. Correlation between PCF and PEmax and VC	The lower limit of effective assisted cough with manual assisted cough, maximum insufflation capacity and maximum insufflation capacity combined with a manual assisted cough was best predicted by VC > 1,030 mL (ROC 0.86, P < .001), VC > 558 mL (ROC 0.92, P < .001), and VC > 340 mL (ROC 0.90, P < .001) respectively. The upper limit of effective manual assisted cough MAC was best predicted by PE_{max} > 34 cm H_2O (ROC 0.89, P < .001). With each of the cough-augmentation techniques the benefits decreased linearly with increasing $PEmax_x$ and VC (P < .001). Patients with SMA Type II had a higher manual assisted cough with a abdominal trust compared to a thoracic manual assisted cough (PCF 245L/min vs. 225L/min (p<0.001)	Societe Francaise de Kinesitherapie	Patients with VC > 340 mL and MEP < 34 cm H(2)O would optimally benefit from the combination of breath-stacking plus manually assisted cough to improve PCF to > 180 L/min however patients were > 8 years old. SMA type II patients achieved a greater improvement in PCF with a abdominal assisted cough. The author recommend the use of MI-E in patient with a VC<340mls.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow -up	Outcomes	Effect size	Funding	Comments
Tzeng, AC. Bach JR.	Prevention of pulmonary morbidity for patients with neuromuscular disease	Cohort	2000 Chest	2-	94, Group 1 n=71, Group 2 n= 23.	68 men, 26 women. Mean age 31, aged 2.5 to 73.5. All had sufficient bulbar function. All home based patients with dedicated family members or personal care attendants.	Patients split in to Group 1: Patients taught maximum insufflaon capacity when VC<2000ml Group 2: Patients with hypoventilation were treated with NIV and manual assisted coughing and maximum insufflations capacity and mechanical insufflation-exsufflation MI-E (+35 to +60 and -35 to -60)	Between group 1 and 2 along with pre and post protocol amount of hospitalisations. Post protocol hospitalisations and avoided hospitalisations.	Study was retrospective.	Hospitalisations of the patient group	Patient groups dependent on ventilator usage None: n=14 Part time: n=73 Full time: n=31 Respiratory episodes/patient/year None:1.06±0.96 Part time: 1.17±1.12 Full time: 0.85±0.87 Hospitalisations/patient/year None:0.04±0.1 Part time: 0.17±0.38 Full time: 0.5±0.12 Hospitalisation avoided/patient/year None:1.026±0.99 Part time: 0.99±1.12 Full time: 0.80±0.85. p<0.001for all groups for episode rate vs. The hospitalisation rate.	A grant from the National Institute on Disability and Rehabilitation Research.	A major limitation of the study is that there is no control group. However, the authors feel this would be unethical. Patients have significantly fewer hospitalisations per year and days per year when using the protocol as needed than without the protocol. The use of inspiratory and expiratory aids can significantly decrease hospitalisation rates for respiratory complications of neuromuscular disease.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Udinkten Cate FEA, van Royen BJ, van Heerde M, Roerdink D, Plotz FB.	Incidence and risk factors of prolonged mechanical ventilation in neuromuscular scoliosis surgery	Retrospective cohort study	2008 Journal of Paediatric Orthopaedics B	3	46	Consecutive children with neuromuscular scoliosis undergoing surgery in a single centre	Scoliosis surgery	A number of pre-operative, peri-operative and post-operative factors were reviewed to assess predictors of post-operative course	Hospital discharge	Prolonged ventilation (>3 days) post-operatively	Those patients with prolonged post-operative ventilation had lower FVC (34% vs 62%) pre-operatively. No other factors were significantly different	Not stated	Only 36 patients undertook lung function testing, and only 6 of these needed prolonged ventilation. Successful extubation was managed in patients with FVC down to 22% predicted.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow -up	Outcomes	Effect size	Funding	Comments
Velasco, M. V., A. A. Colin, D. Zurakowski, B. T. Darras and F. Shapiro	Posterior spinal fusion for scoliosis in Duchenne Muscular Dystrophy diminishes the rate of respiratory decline	Qualitative	2007 Spine	3	56	56 subjects with DMD Mean age 14 yr (range 9.6 – 18.3)	All underwent posterior spinal fusion	Within subject comparison – pre and post op rate of decline of lung function	Up to 8 yr post op	Rate of decline in FVC before and after surgery	Respiratory decline was 4%/yr pre-surgery which decreased to 1.75% post surgery ($p<0.0001$)	No funds was received	Authors concluded posterior fusion for scoliosis in DMD was associated with a sig reduction in the rate of respiratory decline post-surgery compared to pre-surgery. No controls Causal link cannot be confidently established

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Vianello A, Bevilacqua M, Salvador V, Cardaioli C, Vincenti E.	Long-term nasal intermittent positive pressure ventilation in advanced Duchenne's muscular dystrophy.	Qualitative	1994 Chest	3	10	All had Duchenne muscular dystrophy, mean age 20 (12 to 27). All had stable daytime hypercapnia. All were recommended NIV, 5 accepted and 5 declined.	Impact of NIV. Used volume cycled ventilators.	NIV versus standard care.	Followed prospectively for 2 years	Survival, lung function, hospitalisation, blood gases.	After 2 years, 4/5 of patients who declined NIV had died, compared to 0/5 of those using NIV. Of the 4 that died, survival after onset of daytime hypercapnia was 9.7 +/- 5.8 months	Not stated	Uncontrolled study. The 2 groups were not different in severity and the reason for declining NIV was that it was the patient's belief that it would lead to a worse quality of life.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow -up	Outcomes	Effect size	Funding	Comments
Vianello A, Bevilacqua M, Arcaro G, Gallan F, Serra E.	Non-invasive ventilatory approach to treatment of acute respiratory failure in neuromuscular disorders. A comparison with endotracheal intubation	Historically matched cohort study	2000 Intensive Care Medicine	3	14 with 14 historical controls	Adult patients (*-*) with neuropathic disease and acute respiratory failure (ARF)	NIPPV (plus mini-tracheostomy) compared to conventional ventilation	Effect of NIPPV on hospital stay was assessed	Only followed to discharge from ICU	Outcome measures were death or failure of treatment. Length of stay and time to improvement were also compared	Mortality and length of stay was reduced in NIPPV group	Not stated	Small study with historical controls in adult patients. Nevertheless it demonstrates the successful use of NIPPV in patients with NMW.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Vuillerot C., I. Hodgkinson, A. Bissery, A. Schott-Pethlaz, J. Iwaz	Self-perception of quality of life by Adolescents with Neuromuscular diseases	7	2009 J of Adolesc. Health	3	43	Adolescents with neuromuscular diseases mean age 13.8+-1.7yrs 2.9:1 ration male to female	Self administered questionnaire (VSP-A) studying the quality of life	Comparison of results to non-disabled group of age/sex matched healthy adolescents	N/A	VSP-A health-related QuOL questionnaire Motor function measure, physical health assessment Including FVC	Similar in several respects to non-disabled, scored higher in school performance and relationships with teachers. Physical impairment was negatively associated with limitation in activities. Adolescents with ventilator support did not score lower score (67.7+/-11 vs 62.9+/- 15, p=0.39)	AFM (assn. Francaise contre less myopathies) ALLP (Assn Lyonnaise de logistique posthospitaliere)	In this study, the vast majority of disabled adolescents considered their lives worthwhile and enjoyable with surprisingly high QuOL scores.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Wanke, T. Toifl, K. Merkle, M. Formanek, D. Lahrmann, H. Zwick, H.	Inspiratory muscle training in patients with Duchenne muscular dystrophy	RCT	1994 Cheat	1-	30	Patients with DMD (age range 9-24)	strength and endurance inspiratory muscle training (IMT) (10 loaded breathing cycles of one minute duration with 20 seconds between them twice daily and 10 maximal static inspiratory efforts to reach a minimum pressure this was repeated until ten correct attempts had been achieved) for a six month period.	Training versus control. Five of the 15 patients discontinued training after one month as their inspiratory muscle function had not improved. These patients had a severe pulmonary impairment with a VC < 25% predicted and or a PaCO2> 45mmHg. Three of the control group had a decompensation and were excluded from the analysis. There was no significant difference at baseline between the control and patient group.	Patients were assessed 3 months prior to training, at the beginning of training, first, third and end of training. In total 15 months	Training produced a significant increase in maximum oesophageal pressure (Pes_{max}) and maximum transdiaphragmatic pressure (Pdi_{max}) compared to the controls ($p<0.001$).	Endurance training (307.6 ± 126.6 to 448.4 ± 176.7(46% increase)) ($p<0.05$) versus in the placebo group (271.1 ± 40.4 to 271.7 ± 45.6) (p=ns)	Unknown	In DMD the respiratory muscles are trainable in patients with a VC>25% predicted in terms of both strength and endurance.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Ward S, Chatwin M, Heather S, Simonds AK.	Randomised controlled trial of non-invasive ventilation (NIV) for nocturnal hypoventilation in neuromuscular and chest wall disease patients with daytime normocapnia.	RCT	2005 Thorax	1-	26 patients – 12 in the control group and 14 randomised to NIV. Of these 10 in the control group and 12 in the treatment group completed the study, but 9/10 of the controls ended up having treatment	Adults and children with either weakness or scoliosis with nocturnal hypercapnia but not daytime hypercapnia. PLUS a VC<50% OR symptoms of hypercapnia	Nocturnal NIV	Nocturnal NIV or standard care with 6 monthly monitoring	24 months	Peak TcCO2 was the primary outcome measure. Secondary outcome measures were % time above TcCO2 6.5kPa, mean and nadir of nocturnal SaO2, SF36 QOL, lung volumes, cough peak flow, max sniff pressure and max insp and exp mouth pressures	9 out of 10 of controls were started on NIV by 24 months because they reached pre-set criteria for this, most because of development of daytime hypercapnia ($PaCO_2$>6.5 kPa). There was no significant difference in peak TcCO2 at any time point (but even at 6 months, the first evaluation point, 4 of the control patients were already on NIV). Of the secondary outcomes, there was a difference in % nocturnal time>6.5kpa at 6 and 12 months and in mean Sao2. There was a small effect on quality of life in favour of NIV	Not stated	Although this study was planned as an RCT to evaluate benefit of starting NIV on risk of adverse events, its major usefulness is that it demonstrated that once pts have symptomatic nocturnal hypercapnia or nocturnal hypercapnia combined with a VC<50%, they are likely to develop day time hypercapnia within 12-18months. Most of the controls had progressive neuromuscular conditions, and this may not apply to other patient groups.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Winkler, G. Zifko, U. Nader, A. Frank, W. Zwick, H. Toifl, K. Wanke, T.	Dose-dependent effects of inspiratory muscle training in neuromuscular disorders	Qualitative	2002 Muscle and nerve	35	16	Patients (DMD=8, SMA=3) aged 8-29 years old were split into 2 groups those with a decline in VC <10% in the 12 months prior to training (group 1 (n=10) VC mean (SD) 63.23(22.06)% predicted) and those with a decline in VC>10% in the 12 months prior to training (group 2 (n=6) VC 60.20(35.7)% predicted). All patients were stable and did not complain of sleep disturbance, daytime somnolence or morning headaches and had normal daytime ABG's on room air	Strength and endurance training with a device that was constructed to enable patients to adhere to the prescribed intensity.	Comparison was made between group 1 and group 2. Ventilatory function (IVC, FEV1 and 12 sec MVV) and inspiratory muscle function (Pi_{max}) were evaluated 12 months prior to training, at baseline, 1 month, 3 months 6 months and at the end of training (9 months).	2 years	No significant difference in the decline of VC between group I or 2 Improvements in Pi_{max} and 12 sec MVV. Patients with a lower rate of decline of VC (group 1) successfully completed their training programmes and this correlated with improvements in their PI_{max}. Serum creatine kinase was stable prior and throughout the training period	Improvements in Pi_{max} ($p<0.001$) and 12 sec MVV ($p<0.0028$).	Unknown	Strength and endurance training improved Pi_{max} and 12 second MVV respectively. There was no determent to muscle function as monitored by Serum creatine kinase

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Yates K, M Festa, J Gillis, K Waters, K North	Outcome of children with neuromuscular disease admitted to paediatric intensive care	qualitative	2004 Arch Dis Child 89: 170-175	3	28	28 children on 69 admissions (64% male) all acute NM disorders excluded. Median age first admission : 3 yrs 2 mths (range 3mths-19yrs 5 mths)	To determine the outcome of children with neuromuscular disease following admission to PICU (Australia)		July 1986 – June 2001 (15yrs)	Descriptive. Unplanned vs planned admissions. Follow up from 1st admission	16 (57%) had >1 admission. Median stay : 4 days (0.5-42). 23% unplanned resulted in ongoing resp support on discharge. 10(35%) died, 4(14%) in PICU		Most children with NMD admitted to PICU are discharged without the need for prolonged ventilation. But use of home-based NIV is common and likely to require further PICU admission

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Yilmaz O, S A Yildirim, C Oksuz, S Atay, E Turan	Mothers depression and health related quality of life in neuromuscular diseases : Role of functional independence level of the children	qualitative	2010 Pediatrics International	3	40	Children : mean age 8.45yrs+/-3.55(range 3-16) female (n=16) Male (n=24) 26/40 had DMD,8 spinal muscular atrophy,6 had other NMD Primary caregiver was mother, 14/40 received help in care.	Evaluation of relationship between maternal depression and HRQOL and functional limitations of children with NMD.	N/A	N/A	Wee-FIM scores of children's independence Nottingham Health Profile(NHP) Turkish version for mothers HRQOL, Beck depression Inventory (BDI)	Mean total NHP score moderately correlated with total FIM score (p=-0.538) and sphincter control(p=-0.526) weak correlation with locomotion (p=-0.493;P<0.001) moderate correlation between mothers' social isolation and children's sphincter control, social integration and total Wee-FIM (P<0.001)		Functional level of children with NMD affects quality of life of mothers caring for them.

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Young HK, Lowe A, Fitzgerald DA, Seton C, Waters KA, Kenny E, Hynan LS, Iannaccone ST, North KN, Ryan MM.	Outcome of noninvasive ventilation in children with neuromuscular disease.	Qualitative	2007 Neurology	3	14	Children (median age 7.7 years (1.5 to 15)) with neuromuscular weakenss started on NIV – 6 for nocturnal hyopoventilation and 8 following acute respiratory failure	Effect of NIV. All had in hospital physio, but cough assist and other forms of manual cough assist were not used. Home physio is not mentioned.	Hospital admission rate before and after initiation of NIV using hospital records to identify parameters	30 months (6 to 84 months)	Hospital admission and symptoms and health care costs (direct costs associated with outpatient visits and hospital admissions). Did also do QOL but required parents to remember QOL before NIV	Pre NIV 9/14 had daytime sleepiness and 4/14 had headache, compared with 4/14 and 1/14. Pre NIV patients were hospitalized for a mean of 41.7 days/y (range 0 to 82) , after NIV this decreased by 73% to a mean of 10.5 days/y (range 0 to 52) (p =0.002). mean annual hospitalizations in the year before NIV was 3.8/y and was 0.7/y after NIV (p = 0.002). |PICU days decreased from 10.2 days (before NIV) to 2.3 days after NIV (p = 0.06).	Not stated	Uncontrolled study using retrospective data for the pre-NIV events. The hospital admission data is likely to be robust.

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Yousafzai A.K., Filteau S.M., Wirz S.L., Cole T.J.	Comparison of armspan, arm length and tibia length as predictors of actual height of disabled and non-disabled children in Dharavi, Mumbai, India	Case control cross sectional study	2003 Eur J Clin Nutr	3	303 children. Aged 2-6 years	141 with mixed disabilities and 162 non-disabled children.	Height/length, armspan, arm length and tibial length measured to nearest 0.1cm	Relations between armspan, arm length and tibial length with height in the control children were performed using linear regression.	Not applicable	Armspan, arm length and tibial length were found to be strong predictors of height (all $p<0.001$)	Not applicable	Not stated	These measurements can be used to extrapolate a value for height in disabled children who might be non-ambulant or have significant kyphosis and/or scoliosis

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Yuan N, Skaggs DL, Davidson Ward SL, Platzker AC, Keens TG	Preoperative polysomnograms and infant pulmonary function tests do not predict prolonged postoperative mechanical ventilation in children following scoliosis repair	Qualitative	2004 Pediatric Pulmonology	3	110 patients with polysomnography and 18 patients with infant lung function testing	Children and infants (not necessarily with NMW) undergoing scoliosis surgery who were unable to perform lung function testing. Around 50% of the patients had neuromuscular disease (unspecified).	Scoliosis surgery	The incidence of prolonged post-operative ventilation (>3 days)	Hospital discharge		Neither PSG nor infant PFTs were predictive of a prolonged post-operative ventilatory course	Not stated	This was an attempt to improve pre-operative assessment in those patients unable to undertake lung function tests. Only half the patients had neuromuscular disease. The authors note that sleep studies were done during daytime naps and may have missed disturbances related to REM sleep.

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Yuan N, Skaggs DL, Dorey F, Keens TG.	Preoperative predictors of prolonged postoperative mechanical ventilation in children following scoliosis repair	Retrospective cohort /Qualitative	2005 Pediatric Pulmonology	3	125	Patients undergoing scoliosis repair. Of these 57 had neuromuscular scoliosis but this included cerebral palsy and not just NMW	Scoliosis surgery	Demographic, operative and lung function data (VC, FEV1, IC, MIP, TLC and RV)	Hospital discharge	The incidence of prolonged post-operative ventilation (>3 days)	32% patients needed prolonged post-operative ventilation. Factors predictive of this were older patients (OR 1.3), patients with neuromuscular scoliosis (OR 7.7) and those with an FEV1< 40% predicted (OR 8.6). Reduced FEV1 was predictive independent of the diagnosis	Not stated	Only 44% of scoliosis patients were able to perform PFTs. Poor pre-operative lung function was predictive of a prolonged post-operative course. VC< 60% predicted was more sensitive (77%) and FEV1< 40% predicted was more specific (86%). It is unclear whether the inclusion of children with CP influenced the results.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow-up	Outcomes	Effect size	Funding	Comments
Yuan N, C H Wang, A Trela, C T Albanese	Laparoscopic Nissen fundoplcation during gastrostomy tube placement and noninvasive ventilation may ipprove survival in type I and type II spinal muscular atrophy	Qualitative	2007 J of Child Neurol	3	7	6 with SMA1(aged 1.5 to 8 months) and 1 with severe SMA type II (aged 16 mhts) all had respiratory symptoms, difficulty feeding and weight loss. 6 had GO reflux and 5 had NIPPV	Laparoscopic Nissen fundoplication and gastrostomy tube insertion with noninvasive ventilation	Number of pneumonias before and after surgery survival rate compared to determined prognosis	Identified from records from Jan2003 until Aug 2006	number of hospitalizations and pneumonias before and after surgery survival rate (at 1 mth, 3 mths and 12 mths post procedure)	an average of one pneumonia and 1 hospitalization per patient prior to procedure. 1 mth post surgery : total of 1 pneumonia and 1 hospitalization in all patients. 3 mths post surgery (n=6) : average no. of pneumonias and hospitalizations reduced by 50% . 6 mths post surgery : no data due to 6 deaths.	no funding	Consideration of this aggressive surgical treatment even in the absence of symptoms with non invasive ventilation can decrease hospitalization but the medicalisation of feeding and the problems with NIV need balance alongside such a limited life expectancy.

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Yuan N, ane P, Shelton K, Matel J, Becker BC, Moss RB.	Safety, tolerability, and efficacy of high-frequency chest wall oscillation in pediatric patients with cerebral palsy and neuromuscular diseases: an exploratory randomized controlled trial.	RCT	2010 J Child Neurol.	1-	23 participants completed the study. 9 with CP and 14 with NMD	Children with NMD 5 DMD 2 unknown mitochondrial myopathy 2 unknown myopathy 1 CMD 1 SMA type II 3 other or cerebral palsy 10 of the NMD required nocturnal NIV	Effect of HFCWO with the Vest ™	HFCWO vs. conventional physiotherapy	Mean period of 5 months	Adherence to treatment Hospitalisations amount of antibiotics	RCT of HFCWO vs. conventional physiotherapy in patients with NMD and CP. There was no difference between the groups at baseline. Outcomes included a significant improvement with SpO2 post treatment ($p<0.01$) improved adherence ($p<0.036$) decreased but not significant amount of hospital admissions with IVAB ($p<0.09$) for the HFCWO group only	The investigator initiated study was supported in part by a grant from Hill-Rom Inc	No adverse effects were seen in wither the conventional physiotherapy or HFCWO group.

Authors	Title	Study type	Year Journal	Evidence Level	Numbers	Characteristics	Intervention	Comparison	Follow -up	Outcomes	Effect size	Funding	Comments
Zebracki K, D Drotar	Pain and activity limitations in children with Duchenne or Becker muscular dystrophy	qualitative	2008 Dev Med and Child Neurol	3	53	53parent/child dyads. DMD n=43, aged 13.88yrs (SD 3.38); BMD, n=10, aged 14.8yrs (SD 1.48). 79.1%DMD non-ambulant, 5(50%) non-ambulant. Surgery for DMD(n=26), BMD (n=5)	To examine prevalence and characteristics of pain in DMD/BMD, including nature of disagreements between parents, physicians and limitations on daily activities		N/A	Pain intensity, frequency , location and distress (rating scales, body map) Child Activity Limitations Interview	Self reported pain : 54-80%, parent reports 70-90%. (Agreement poor to fair)Typically once weekly, mild to moderate intensity. Particularly lower back, spine and legs	National Institute of Mental Health Pediatric psychology Research Training Grant.	Pain is common and under recognized. Pain assessment needs to be part of standardised care