Indicators of Need for Mechanical Ventilation in Duchenne Muscular Dystrophy and Spinal Muscular Atrophy

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Study objectives: **The purpose was to investigate a possible relationship between different parameters of physical function, spirometric measurements, and the approaching need for mechanical ventilation.**
Design: **A nonrandomized, prospective, descriptive study of 11 patients with spinal muscular atrophy type II (SMA-II) and 14 patients with Duchenne muscular dystrophy (DMD). At a home visit, the anthropometric indices of age, height, and weight were recorded, the degree of disability was scored, and measurement of the strength of eight muscle groups and spirometry was performed. The interdependence of the variables was analyzed and the intergroup differences evaluated. Eighteen months later, it was found that one of the authors (B.J.), who was blind to the results of the first examination had instituted home mechanical ventilation on seven of the patients. The data were analyzed retrospectively for their predictive value as indicators of approaching ventilator dependency.**
Results: **The seven patients who needed mechanical ventilation were the patients with DMD with the highest disability score (Egen Klassifikation [EK] sum >20) and the smallest values for FVC <1.2 L (FVC% <30). We found a significant correlation (p=0.002) between FVC% and the EK sum at the first examination and between the FVC% and the time until treatment with mechanical ventilation was instituted (p=0.023). Although 7 of the 11 patients with SMA type II had FVC below 1.2 L and some of them had an EK sum score higher (indicating more disability) than some patients with DMD who needed mechanical ventilation, none of them required mechanical ventilation.**
Conclusion: **In this investigation, a combination of EK sum and FVC% provided a better indication of the approaching need for mechanical ventilation in the patients with DMD than the variables separately.**
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CPAP=continuous positive airway pressure; DMD= Duchenne muscular dystrophy; EAMDA=European Alliance of Muscular Dystrophy Associations; EK=Egen klassifikation; FEV_1=forced expiratory volume in 1 s; FVC=forced vital capacity; PEF=peak expiratory flow; SMA II=spinal muscular atrophy type II

Key words: disability score; home care mechanical ventilation; lung function; neuromuscular disease

The precise time for instituting mechanical ventilation in patients with neuromuscular disease presents the clinician with a dilemma unless the patient is admitted to the hospital in a life-threatening respiratory crises. However, decisions made during an acute episode of respiratory insufficiency are often less than ideal and it is our experience that patients with different types of muscular dystrophy are being treated more and more often with elective mechanical ventilation at home.[1] The study reported herein is concerned with determining clinical indicators at the stage of the disease at which there is impending daytime hypoxemia that will require full ventilatory support.

Duchenne muscular dystrophy (DMD) and spinal muscular atrophy type II (SMA-II) are both genetically determined neuromuscular diseases characterized by profound global skeletal muscle weakness. In DMD, the disease is classically described as involving the proximal muscle groups with progressive loss of strength so that the ability to ambulate independently is lost at around 9 years of age. Although studies have shown that involvement of the muscles of respiration is already advanced at this stage and that on formal lung function testing boys with DMD have markedly reduced vital capacity, it is not usually until the stage of wheelchair dependency that the loss of respiratory function becomes a major complaint. In subsequent years, the reduction in respiratory competence becomes dominant and according to one report responsible for death in 90% of patients.[2]

In SMA type II, which is generally considered to be a nonprogressive neuromuscular disease, arising from the destruction of the anterior horn cells, the muscle weakness is of such magnitude that the child may never have attained the motor milestones of standing and

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walking. Characteristically, onset is at or around the time of birth and they have marked weakness of the trunk and anterior abdominal muscles often associated with varying degrees of hypotonicity.

In clinical practice, three stages are seen in the decline of respiratory competence in patients with DMD, namely, (1) difficulty in clearing secretions and microatelectasis; (2) nocturnal hypoventilation; and (3) daytime hypoxemia.

In Denmark, regular but intermittent use of continuous positive airway pressure (CPAP) has been found to be beneficial in relieving the signs and symptoms associated with the first stage. Noninvasive ventilator support is used to manage nocturnal hypoventilation, which is identified on the basis of changes in oxygen and carbon dioxide blood gas levels during sleep studies. However indicators for making the transition to full ventilator support are less clear. The decision is compounded by the fact that many patients do not obviously pass through the second stage or it remains unrecognized.

Improved home management techniques for the ventilator-dependent patient, technologic advances in the design of ventilators, and the wider acceptance of home ventilator support as appropriate management make it essential to attempt to determine simple clinical indicators for impending ventilator dependency.

The purpose of this study is to evaluate various clinical parameters as possible predictors for the need for future mechanical ventilation in patients with DMD and SMA-II.

METHODS

Study Design

This was a descriptive, nonrandomized study. All 43 patients, all from Jutland, Denmark, registered at the Treatment and Guidance Centre for the Muscular Dystrophy Association with the diagnosis DMD or SMA-II, without tracheostomy or using any form of mechanical ventilation, were asked to participate in a study to monitor respiratory function and disability level. The project had been accepted by the Ethical Committee, Århus, Denmark, and informed consent was obtained from all subjects.

The patients were to be examined and evaluated in their homes by two of us (B.S. and S.L.) using the method described. One of us (B.J.) was blinded to the results but was the clinician responsible for evaluating the patient for mechanical ventilator support, if, he or she was admitted to hospital in respiratory failure. Eighteen months after the start of the study, we found that seven of the patients had required mechanical ventilation.

Subjects

Of the 43 persons who were invited to participate, 4 refused, 14 did not reply, and 25 accepted; of those accepting, 14 were boys with DMD and 11 were patients with SMA-II. Their diagnosis had been established in neurology departments in hospitals throughout Denmark. All the patients were wheelchair bound and had never suffered overt respiratory insufficiency or seriously discussed the need for mechanical ventilation. All participants used CPAP apparatus daily for pulmonary hygiene and were under supervision from the Treatment and Guidance Centre, Århus.

All patients were assessed with respect to respiratory competence and physical function. The following clinical parameters were obtained and measured: (1) anthropometric data, age, sex, weight, and arm span (the maximal horizontal distance between the fingertips of the two hands);[3] (2) measurement of spirometric indices of pulmonary mechanics; (3) evaluation of the level of functional disability; and (4) measurement of the strength of eight muscle groups.

Spirometric Indices of Pulmonary Mechanics

Measurement of forced vital capacity (FVC), forced expiratory volume in 1 s (FEV_1), and peak expiratory flow (PEF) were made, using a calibrated vitalograph (COMPACT; Vitalograph Ltd; Buckingham, UK). From the FVC and FEV_1, the ratio of FEV_1/FVC was calculated. The subjects were all measured in the sitting position and in addition, 12 of them (6 with DMD and 6 with SMA-II) were randomly selected and also measured in the supine position.

A nose clip was always used and special attention was given to keeping the connection between the lips and the mouthpiece tight during measurements. The measurements were done during a forced expiration from the total lung capacity to the residual volume and recorded as flow-volume curves with flow rate (Liter·min^{-1}) against expired volume (Liter). The best attempt out of three for FVC was recorded and used as the basis for the calculations on each of three occasions over a 4-h period.

All measured volumes were recalculated to body temperature and pressure saturated conditions, *ie*, 37°C, saturated with water vapor. All measured indices were compared with the individually calculated reference values[4] for FVC%, FEV_1%, and PEF%. Calculating the reference value, we used the sex, age, and the maximal horizontal span between the finger tips from the right to the left hand, the latter being a proxy for height. Studies have shown that in normal healthy humans, there is not more than 5 cm difference between height and arm span.[3] We did not use height because some of these patients had marked scoliosis.

The coefficient of maximal variation on double determinations, greatest and smallest value for each patient, was FVC of 4.0%, FEV_1 of 4.0%, and PEF of 9.2%.

Classification of Disability

Two methods were used to evaluate the subjects' level of disability: the Egen Klassifikation (EK) sum (B. Steffensen, SRP, personal communication, January 1990) and the disability classification proposed by the European Alliance of Muscular Dystrophy Associations (EAMDA) classification or scoring systems.[5] The EK is determined by use of a questionnaire that covers ten main competencies of the ability to do the following: (1) control an electric wheelchair; (2) transfer from the wheelchair; (3) stand; (4) sit up; (5) use the arms (generally); and (6) use the arms for eating; (7) turn in bed; (8) cough; (9) talk; and (10) general well-being that is judged from the statements made by the patients about headache, feeling of exhaustion, sudden changes in mood, disturbed sleep, or nightmares. Each of the main issues is scored from 0 to 3, in which 0 indicates the best possible functional level for a patient with DMD who has lost the ability to walk independently and in which 3 indicates that the patient cannot move without help. The total score (EK sum) ranges between 0, representing maximal independence in the wheelchair, and 30 signifying total dependence on other people's help.

The EK was designed by one of the authors (B.S.) as a scale reflecting the progressive loss of physical ability in patients with DMD, but was used in this study also for patients with SMA-II. A comprehensive description of EK can be requested from the authors.

The EAMDA classification is divided into five main areas: (1) general well-being, described by 11 different questions, which can be answered yes or no; (2) dyspnea; (3) speech; (4) coughing; and (5) swallowing. Areas 2 to 5 are divided in four or five levels (0 to

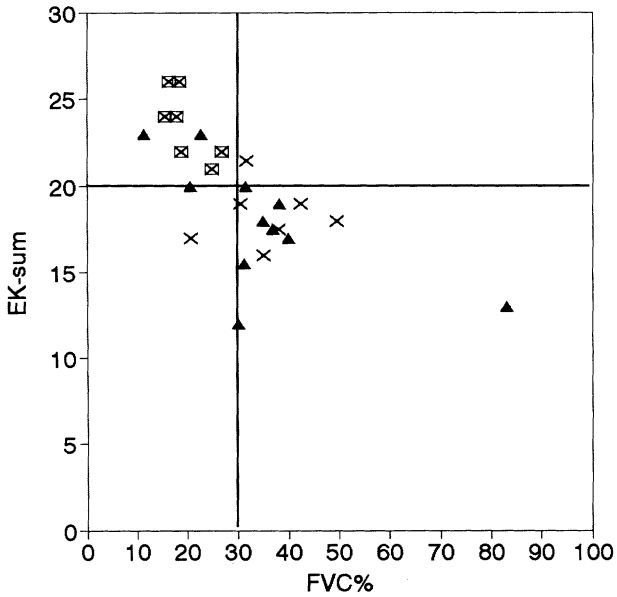

FIGURE 1. Relationship between disability score and the forced vital capacity in percent of measured value (FVC%) in the DMD and the SMA-II groups. EK sum vs FVC% for the patients with DMD (x) and the patients with SMA-II (triangles). Those who were later mechanically ventilated are identified by boxes. The vertical line indicates FVC%=30%; horizontal line indicates EK sum=20.

4 or 5) describing increasing disability. The individual scores were summed and are herein called the EAMDA sum. The range was between 5, maximal independence, and 30, totally reliant on help.

Muscle Strength

Measurement of the voluntary muscle strength of eight muscle groups, namely the right and left elbow flexors and extensors and the knee flexors and extensors were made using a hand-held dynamometer with a range of 0 to 30 kg (myometer; Penny & Giles; Dorset, UK). The values obtained for all eight muscle groups were summed. The measurement was always made by the same author (B.S.).

Procedure for Institution of Mechanical Ventilation

It is standard practice for all patients from Jutland with a neuromuscular disease and incipient respiratory insufficiency to be transferred to the Danish Respiratory Centre West from neurologic departments, intensive care units, or general practitioners for evaluation. During 3 or 4 days of hospitalization, they undergo a normal clinical examination, radiography of the thorax, electrocardiography, echocardiography, nocturnal transcutaneous measurement of oxygen and carbon dioxide tensions and oxygen saturation, and an estimation of electrolytes in blood and urine. If signs and symptoms reveal respiratory insufficiency to a degree that would normally make mechanical ventilation necessary, the patient is offered this treatment. No systematic measurement of lung volumes is done at that time.

Statistical Treatment

The small numbers of subjects in the study groups and the ordinal or ranking scale character of many of the measurements made the use of nonparametric methods appropriate. The figures are presented as medians with ranges, the two groups are compared with two-sample rank sum test (Mann-Whitney test), and the correlations are done with a Spearman Rank test. Accepted level of significance is $p \leq 0.05$ (two-tailed test).

RESULTS

The results obtained for all parameters for the DMD group and the SMA-II group are given in Table 1, where medians and ranges are shown for each group and the significance levels on the comparisons of the two groups are reported.

There were no statistically significant differences in any of the measured parameters between the five female and the six male subjects among the patients with SMA-II. Therefore these patients are considered as one group.

Anthropometric Data

As can be seen in Table 1, there were large differences between subjects, but not between groups for the anthropometric measures of age and the ratio arm span/weight. The DMD group as a whole was heavier than the SMA-II group.

Spirometric Indices of Pulmonary Mechanics

The median values obtained for the spirometric measurements FVC, FEV_1, and PEF for both groups are given in Table 1. There was no significant difference between the two groups of patients in any of these indices. In the SMA-II group, one patient could not perform the PEF test. Of the 12 patients randomly selected to be measured in the supine position as well as in the sitting position, the six patients with SMA-II had a substantially higher FVC in the supine position than in the sitting position. The increments had a median value of +0.220 L (range +0.130 to +0.680 L). The FVC increased to a value larger than 1.2 L; actual value was 0.95 to 1.45 L in only one patient. Among the six patients with DMD only one demonstrated an incre-

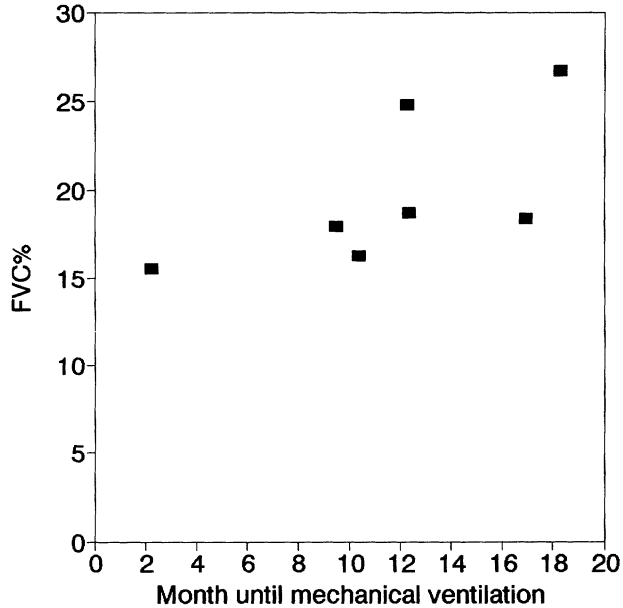

FIGURE 2. FVC% vs time span (in months) between our examination and the start of mechanical ventilation.

Table 1—*Summary of Data for all Parameters Measured for Both Groups of Patients, Analyzed for Comparison

	DMD (n=14) Median (Range)	SMA-II (n=11) Median (Range)	p Value
Age, yr	19.2 (15.4-30.5)	25.8 (9.3-52.5)	0.13 (NS)
Weight, kg	61 (38-90)	40 (19-67)	0.03
Span, cm	167 (154-190)	150 (132-170)	0.01
Weight/span	0.40 (0.22-0.53)	0.27 (0.14-0.40)	0.08 (NS)
FVC, L	1.11 (0.73-2.18)	1.06 (0.27-3.89)	0.54 (NS)
FVC%	25.8 (15.6-49.5)	31.5 (11.3-82.8)	0.32 (NS)
FEV_1%	28.5 (17.4-51.2)	31.6 (22.4-87.0)	0.26 (NS)
PEF%	34.9 (24.4-57.2)	41.6 (23.7-96.6)	0.89 (NS)
EK sum	21.3 (16-26)	18.0 (12-23)	0.09 (NS)
EAMDA sum	6.8 (5-15)	6.0 (5-13)	0.43 (NS)
Force, UE	2.6 (0.8-6.3)	1.9 (1.0-5.8)	0.89 (NS)
Force, LE	3.5 (1.2-10.1)	2.4 (1.1-5.1)	0.24 (NS)
Force, sum	6.9 (2.0-16.0)	4.3 (2.12-10.4)	0.44 (NS)

*Differences between DMD and SMA-II. The parameters are tested with a Mann-Whitney rank sum test. One patient with SMA-II could not perform the PEF test (n=10). NS=Non significant (p>0.05); span=the maximal distance (in centimeters) between the fingertips on the right and left hands; FVC%=FVC in percent of calculated reference value; FEV_1%=FEV_1 in percent of the calculated reference value; PEF%=PEF in percent of the calculated reference value; EK sum=disability scoring system (authors' design); EAMDA sum=disability scoring system proposed by EAMDA; Force=muscle force (kilograms, kg); UE=sum of four muscles in the upper extremities (kilograms, kg); LE=sum of four muscles in the lower extremities; sum=the sum of the force in all eight muscles (kilograms).

ment in FVC from the sitting to supine position. The median value was 0.045 L, virtually zero since the range was from +0.18 to −0.14.

Disability Scores and Muscle Strength

The results of the two groups of subjects for disability level using the EK sum and EAMDA assessments are given in Table 1 together with those for summed muscle strength. It can be seen that there were no significant differences between the two groups for any of these parameters.

Relationship Between Disability Scores and Pulmonary Mechanics

To explore the covariation between the disability level and each of the six pulmonary function indices, Spearman rank correlation tests were done. Using the EK sum as the measure of disability, significant correlations were found with all the six indices of lung mechanics measured in the DMD group, four reaching statistical significance at the p<0.01 level. However, when the EAMDA sum was used as the measure of disability, a much weaker relationship was found among these variables in the same group, only four indices achieving statistical significance at the p<0.05 level, the remaining two being nonsignificant. To illustrate the different level of covariation, in the DMD group, EK sum vs FVC% had a Spearman's r(S)=−0.752 (p<0.002), while the same analysis using the EAMDA disability level produced r(S)=−0.645 (p=0.013). This finding encouraged a second-level analysis of the data. The last three competencies, 8, 9, and 10 of the EK assessment, are directly comparable with those areas assessed in the EAMDA system, and were used as a subset (EK 3). The remaining seven competencies (EK 7) provide supplementary information on the mobility aspects of physical function. Spearman rank correlation tests using the two subsets vs FVC% were repeated. Although there was still a significant correlation (p<0.05) for both subsets, it was not as strong as using the EK sum.

In the SMA-II group, there was a weak correlation with EK sum in only two of six indices (p<0.05) and no statistically significant relationship with the EAMDA sum, and when the analysis was repeated using the subsets, the correlation was much less evident, reflecting the greater range in pulmonary indices seen in these patients.

The patients with the smallest values for the pulmonary indices have the highest disability scores. Figure 1 shows the relationship between the disability score obtained by the EK sum method and the FVC% for the two groups. The individual results for each patient are plotted in X–Y diagrams with the FVC% on the X-axis and the related EK sum on the Y-axis.

Patients Who Required Mechanical Ventilation

Signs and symptoms from the seven patients with DMD at the time when mechanical ventilation in the home was instituted are shown in Table 2. The information on case 14 is missing. He had a respiratory arrest during a visit to the cinema. He was resuscitated and this was followed by mechanical ventilation. After unsuccessful attempts to wean him from the ventilator, he was transferred to the respiratory unit. The other six patients were hypercapnic during the day (Table 2). They were offered a tracheostomy and a respirator (type PLV 100). One of them (case 13) would not accept a tracheostomy, and he has, to date, been treated with nasal-mask-ventilation by a ventilatory support

Table 2—*Signs and Symptoms Precipitating the Start of Mechanical Ventilation

	Case No.						
	7	12	13	14	20	22	23
Accumulation of secretions	+	+	+	?	+	+	+
Fits of choking	+	+	+	?	–	+	+
Recurrent infections of lung/airway	–	–	+	?	+	+	+
Hypercapnia, night/day	+/+	+/+	+/+	?	+/+	+/+	+/+
$PaCO_2$ during day, kPa	6.3	6.9	6.7	?	7.4	7.9	8.3
Hypoxemia, night/day	+/–	+/–	+/–	?	+/–	–/–	–/–
Night sweats	+	+	+	?	+	–	–
Disturbed sleep	+	+	+	?	+	+	+
Attacks of fear/anxiety	–	+	+	?	–	+	+
Nightmare	–	+	+	?	–	–	–
Morning headache	–	+	+	?	+	+	–
Slow awakening	–	+	+	?	+	+	–
Epigastric oppression	–	+	+	?	+	–	–

*Plus sign=presence of symptoms/signs; minus sign=absence of symptoms/signs; question mark=not known; hypercapnia=increased P_{CO_2} in samples of arterial blood; hypoxemia=decreased P_{O_2} in samples of arterial blood.

system (BiPAP; Respironics Inc; Murrysville, Pa.)

The seven patients with DMD who needed mechanical ventilation are included in Figure 1, and it can be seen that these seven patients are characterized by having an FVC <1.2 L, corresponding to FVC% <30, and EK sum >21. They had FEV_1 <1.11 (FEV% <30), PEF <200 L min^{-1} (PEF% <38), and EAMDA sum ≥6.

One patient, case 19, fulfilled the same criteria for spirometric tests (FVC%=20.1) without needing a ventilator, but his EK sum score was only 17. Another patient, case 17, had an EK sum score as high as 22, but his FVC% was 32% and he did not need mechanical ventilation. In Table 3, the subgroup comprising those patients who later went on to use ventilators are compared with those subjects with DMD who did not. There was no statistical difference in relation to age or muscle force (UE, LE, or sum [see Table 1 footnotes for explanation]) between the subgroups. However, as shown, there were highly significant differences in all the measured indices for pulmonary function and the disability scores with the EK sum achieving a significance level of $p<0.001$. Figure 2 shows the relationship between the FVC%, measured in the home on entry to the study, and the time interval in months until treatment with mechanical ventilation was established. The correlation was significant: Spearman rank correlation r(S)= 0.821, p=0.023.

There were no other significant correlations between this time interval and anthropometric data, pulmonary indices, disability scores, or muscle force.

As shown in Figure 1, three patients in the SMA-II group had FVC% less than 30% and EK sum scores of 20 and above, but none of these patients required ventilation.

Discussion

In patients with lung diseases such as COPD, alteration in lung volumes primarily reflects changes in the lung parenchyma and airways. In patients with neuromuscular diseases, the measured lung volumes are largely determined by the subject's muscle strength and if spinal deformity is present, decreased chest wall compliance producing a secondary restrictive effect on the lungs. Clinically, FVC may be said to be a proxy for a muscle test for the total effect of the respiratory muscles. Although measurements of peak expiratory mouth pressure and peak inspiratory mouth pressure[6] provide more specific information on respiratory muscle weakness, they do not completely describe the mechanical impairment in the two groups studied.

Forced vital capacity is very reproducible, even in patients with neuromuscular diseases; in this investigation, the coefficient of maximal variation was only 4%. This makes it applicable to sequential investigations of changes in the spirometric values of lung mechanics.

The maximal horizontal span between the finger tips from right to left hand was used as a substitute for height in this study, a method that has been shown to be within 5 cm of height in normal subjects. However, we are aware that some researchers have reported that this is an unreliable method in patients who have shoulder, elbow, or wrist contractures and could reduce the accuracy, so influencing the predicted normal spirometric volumes.[7] However, the patients in our study groups had only minimal contractures.

The FVC and other force-dependent indices for pulmonary mechanics are probably important in relation to the patients ability to cough and clear secretions from the airways. Its relationship to elimination of carbon dioxide and uptake of oxygen remains unclear

Table 3—*Comparison of Those Patients With DMD Who Became Ventilator Users With Those Who Remained Without a Ventilator**

	DMD (+ R) (n=7) Median (Range)	DMD (− R) (n=7) Median (Range)	p Value
Age, yr	19.5 (15.4-30.5)	18.8 (16.2-29.69)	1.000 (NS)
Weight, kg	56 (38-65)	64 (50-90)	0.017
Span, cm	163 (154-176)	169 (160-190)	0.165 (NS)
Weight/span	0.29 (0.22-0.38)	0.37 (0.30-0.53)	0.053 (NS)
FVC, Litre	0.84 (0.73-1.13)	1.78 (1.08-2.18)	0.002
FVC%	18.4 (15.6-26.8)	35.1 (20.8-49.5)	0.002
FEV_1%	18.7 (17.4-29.5)	35.1 (23.9-51.1)	0.002
PEF%	33.0 (24.4-37.69)	48.4 (33.8-57.2)	0.011
EK sum	24.0 (21-26)	18.0 (16-21)	0.001
EAMDA sum	8.5 (6-15)	6.0 (5-8.5)	0.017
Force, UE	1.4 (0.8-3.7)	3.6 (1.25-6.3)	0.209 (NS)
Force, LE	2.7 (1.2-6.4)	5.3 (1.2-10.1)	0.318 (NS)
Force, sum	4.4 (2.0-9.2)	7.8 (2.5-16.0)	0.259 (NS)

*DMD (+ R)=the later ventilator users compared with those with DMD who did not require ventilator support, DMD (− R); NS=p>0.05. See Table 1 for explanation of abbreviations.

in neuromuscular disease. One study reported that despite significant respiratory weakness, only 7 of 16 patients with DMD demonstrated a $PaCO_2$ >45 mm Hg, but they found a significant correlation between FVC% and $PaCO_2$.[8]

Other studies[9] report similar findings, and in DMD, because the patients experience chronic hypercapnia, blood gas levels are probably more important criteria in determining the time at which to start nocturnal ventilation rather than continuous ventilation. The patients in our study group all experienced respiratory failure during relatively minor insults to the respiratory system. It could be postulated that the weakness of the muscles of respiration had reached the point where they were no longer able to respond to even small increases in work load and that the compensatory mechanisms for adapting to chronic hypercapnia were exhausted and therefore respiratory collapse was sudden.

On the basis of our findings, an evaluation of FVC% or FVC alone is not sufficient to predict impending respiratory insufficiency in patients with SMA or DMD. The seven patients with DMD who required mechanical ventilation had an FVC% below 30, EK sum >21, and EAMDA sum >6. However, one patient with DMD, case 19, had FVC% of 20.1, without needing ventilator support, but this patient had retained a high level of mobility. Although there was no significant correlation individually between EK sum and EAMDA sum and the time span to mechanical ventilation combining one criterion of pulmonary function, FVC% <30% with the criterion EK sum >20, we could have identified at the time of entry to the study all those patients with DMD who later needed mechanical ventilation. None of the patients with SMA-II needed ventilation, although seven of them had an FVC <1.2 L, and three had FVC% <30% (Fig 1); but again, these patients showed a trend to retain greater degrees of mobility on disability evaluation.

The information on accumulation of secretion, symptoms of choking, problems with swallowing and coughing, disturbed speech, and symptoms of hypercapnia and hypoxemia is scored by both EK and the EAMDA disability assessments. Thus, one might expect a relationship between the disability scores and the approaching need for mechanical ventilation. However the EK sum also assesses the patient's ability to move and this is of importance in relation to competence in clearing secretions and as an indirect method of assessing muscle strength at a stage in the course of the disease when formal muscle testing would not yield pertinent information. The very low values obtained for the eight muscle groups tested in this study are indicative of the profound muscle weakness.

In the SMA-II group, seven patients had disability scores which, in agreement with the criteria described above, should indicate a need for mechanical ventilator support, but none of them required it. The reasons that patients with SMA-II do not follow this pattern remain unanswered. The explanation might be that the function of the diaphragm is better preserved in patients with SMA-II than in patients with DMD.[10] The patients with SMA-II who in this investigation had the FVC measured in the supine position showed an increase in FVC in the supine position. One possible explanation is that because the diaphragm is pushed upwards into the thorax and the muscle fibers are stretched when the patient is lying down, the patients with SMA-II have a better ventilation during rest than the patients with DMD.

Other possible mechanisms are that the larger vital capacity in supine position is related to a reduction in residual volume rather than to an increased mechan-

ical advantage of the diaphragm, as a similar finding was reported in tetraplegic patients.[11] Further, a study[12] of long-term ventilatory support in patients with SMA reported that institution of mechanical ventilation in those subjects was not required until the FVC was only 20% of the predicted value and none of the subjects in the present study declined to this value.

Lastly there is evidence to suggest that failure of intercostal muscle tone to stabilize the chest wall results in a breathing pattern in which there is inward distortion of the rib cage during inspiration; in this case, diaphragmatic movements create enough intrathoracic pressure during inspiration to cause loss of rib-cage volume.[13]

The precise indications for starting home mechanical ventilation in patients with neuromuscular diseases are as yet undetermined.

The motivation of the patient for seeking help and accepting the treatment is, of course, predominantly related to the symptoms (Table 2). From the patient's point of view, the success of the treatment is also related to a reduction or elimination of the symptoms.

Another factor that becomes increasingly obvious from the literature is that patient acceptance of ventilatory support has increased with the technological advances in the methods of delivering ventilation. The availability of noninvasive techniques such as nasal masks and nasal prongs in combination with smaller mechanical ventilators for home use means that quality of life can be preserved and that the introduction of full-time ventilator support can be introduced in stages. However, the patients who were ventilated in this study were not suitable for noninvasive mechanical ventilation using a BiPAP system, as they were hypercapnic during both the day and night. Also the relatively active and aggressive lifestyle favored by the patients makes tracheostomy more practical.

We recognize that this study is limited by the small numbers and that the low uptake (25 of 43) might be considered to have introduced bias into the findings, but it is also possible that this small group of 43 patients is "overstudied," and was too actively engaged in an extrovert lifestyle to want to participate in our study.

Our observations suggest that by using a simple clinical test for FVC in conjunction with the EK disability scoring system, it is possible to identify patients with DMD who are most at risk of impending respiratory insufficiency. Further, these tests are easily undertaken in the home by paramedical staff and therefore offer a cost-effective method of monitoring the at-risk group.

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