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FUNCTIONAL STATUS AND MUSCLE STRENGTH IN PEOPLE WITH DUCHENNE MUSCULAR DYSTROPHY LIVING IN THE COMMUNITY

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Objective: **To describe activity limitation of people with Duchenne muscular dystrophy who are living in the community and to correlate it with age and muscle strength.**
Design: **Descriptive, correlational.**
Subjects: **Twenty-seven children with Duchenne muscular dystrophy aged 7–14 years who are living in the community.**
Methods: **The subjects' activity limitation was evaluated using the Functional Independence Measure and the muscle strength of their major upper and lower limb muscles was evaluated with manual muscle testing. The Functional Independence Measure was correlated with age and manual muscle testing, and the pattern of activities of daily living limitations and factors related to it were analysed.**
Results: **There were significant correlations between age and averaged MMT score (Spearman's rho = −0.63, $p < 0.01$), age and Functional Independence Measure motor score (rho = −0.52, $p < 0.01$), and Functional Independence Measure motor score and averaged manual muscle testing (rho = 0.77, $p < 0.01$). At similar manual muscle testing level, children with good cognitive function (Functional Independence Measure cognitive score ≥26) showed significantly higher Functional Independence Measure motor scores than those with poor cognitive function (Mann-Whitney U test, $p < 0.01$). For individual Functional Independence Measure items, eating and bowel management were the easier, whereas transfer and stair climbing were the more difficult. Patients with mean muscle strength ≥grade 3 were rated as relatively independent, while those with a mean muscle strength <grade 3 were rated as maximal or total assistance (Mann-Whitney U test, $p < 0.05$).**
Conclusion: **Activities of daily living in patients with Duchenne muscular dystrophy are related to age and muscle strength, and manual muscle testing grade 3 is an important cut-off point to predict their disability.**

Key words: neuromuscular disease, muscle weakness, functional instrument, rehabilitation.

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INTRODUCTION

Duchenne muscular dystrophy (DMD) is a genetic disorder of childhood characterized by lack of the protein dystrophin in muscle membranes (1). It demonstrates a rather uniform progression of skeletal muscular weakness and functional deterioration (2). As the disease progresses, patients become more and more dependent in activities of daily living (ADL) and the care load increases progressively. To provide comprehensive rehabilitative management to maximize their function and quality of life (QOL) it is essential to assess patient's disability objectively, and to determine the factors related to it.

There have been, however, limited studies assessing these patients' ADL with standardized instruments in the literature. Lue et al. (3) studied deficits of ADL skills with the Barthel index (4) in 35 patients with DMD and found that most assistance was required with bathing and least with feeding. With multiple regression analysis, they concluded that hip contracture and static sitting balance were important factors influencing ADL. Nair et al. (5) quantified disabilities in 31 children with DMD with the Barthel index, and reported that among the impairment variables, muscle strength correlated with the Barthel index score.

In Japan, approximately half of the estimated total of 4000 people with DMD have traditionally been cared for in government-supported long-term care facilities (6). Their ADL has been studied with an ADL scale developed by the DMD study group funded by the Ministry of Health and Welfare (currently the Ministry of Health, Labour and Welfare) and several reports are available in Japanese literature describing their ADL (7, 8). The reliability and validity of the instrument used, however, have not been formally tested. Furthermore, data are limited to institutionalized patients, and information is lacking for children living in the community. With the proliferation of the idea of normalization and the more recent emphasis on functioning and participation as proposed in the International Classification of Functioning, Disability and Health (ICF) (9), more and more people with DMD are living in the community in Japan. Because the living conditions in the community are much more diverse than in institutions, it is important to describe patients' ADL with a standardized instrument to plan appropriate rehabilitative management.

The Functional Independence Measure (FIM^{SM}) is a standardized measure of disability with well-documented psychometric

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properties (10), and has been used widely internationally in medical rehabilitation of patients with stroke (11), traumatic brain injury (12), spinal cord injury (13) and so on. It consists of 18 items, organized into 6 categories: self-care, sphincter control, mobility, locomotion, communication and social cognition. It is rated from 1 (complete dependence) to 7 (complete independence) and the total score ranges from 18 to 126, the higher scores indicating less severe disability. It is considered more sensitive for detecting deficits and changes in ADL than the Barthel index, which is a 3-point scale. It is potentially a useful instrument for describing disabilities in persons with DMD, but to our knowledge, there has been no published study assessing their ADL in detail with the FIM^{SM}. The purpose of our study is to quantify ADL in people with DMD living in the community using the FIM^{SM}, and to relate it to age and muscle strength of major limb muscles.

METHODS

This study was performed at a muscular dystrophy clinic of a regional centre for neuromuscular diseases in Saitama Prefecture, in the northern part of the Tokyo Metropolitan Area with a population of approximately 7 million in 2002, where the total number of patients with DMD is estimated as approximately 240 based on the incidence of DMD (6.3/100,000 live births) and the number of males (3.5 million) in the prefecture (14). Among them, 126 patients are regularly followed-up at the clinic, and inpatient care is provided for 66 patients who are older (mean age 25.3 years) at the centre. For this study, we selected patients living in the community aged 7–14 years for the following reasons: (i) the focus was on ADL of people living in the community; (ii) manual muscle testing is difficult to perform reliably in younger patients (15); (iii) because loss of ambulation is known to occur at about 9–11 years and the ability to sit independently is lost at about 15 years (2), we assumed that in this age range, we could observe a wider spectrum of ADL abilities; (iv) McDonald et al. (16) reported that there was a rapid decline in strength between the ages of 5 and 13 years, but after the age of 13 years, the decline was less. (v) The analysis of function in the very young child would be confounded by expected developmental pattern. All these considerations implied that the age range we selected was likely to be a period of rapid change in ADL.

We recruited 27 consecutive patients aged 7–14 years who visited our muscular dystrophy clinic between October 2001 and May 2002. No patients had acute illness. All of them were males, and their mean age was 9.7 years (SD 2.0). The diagnosis of DMD was confirmed by gene analysis. The purpose and procedures of the study were fully explained and informed consent was obtained from their guardians.

One of the authors, well-trained in the use of the FIM^{SM}, assessed their ADL with it by interviewing their parents. We studied the relation between FIM^{SM} scores (motor and cognitive scores and individual item scores) and age with a Spearman's rank correlation method (17). For each FIM^{SM} item, we also calculated the percentage of patients requiring assistance (FIM^{SM} score <5), supervision (FIM^{SM} score $= 5$) and patients who are independent (FIM^{SM} score ≥ 6) to indicate item difficulty.

In addition, a physiatrist experienced in DMD rehabilitation graded their muscle strength according to the Medical Research Council (MRC) grades (18) for flexors and extensors of shoulder, elbow, wrist, hip and knee joints and dorsiflexors and plantarflexors of ankle joints bilaterally. Although no formal reliability testing of MMT was performed for this study, the measurement was carried out according to a manual specifically developed for DMD population by a national DMD study group to assure reliability of MMT (19).

We averaged MMT scores of the upper, lower and all 4 limb muscles as described by Fowler et al. (20), and investigated the relationship between FIM^{SM} scores and the averaged strengths with the Spearman's rank correlation method. Furthermore, we divided the patients into subgroups as defined by the FIM^{SM} cognitive score (>25, i.e. independent, or ≤ 25, i.e. requiring supervision or assistance) or MMT level (<3 or $3\leq$) and analysed the differences in FIM^{SM} motor scores between these subgroups.

All the analyses were performed with Statcel^{TR} (OMS, Saitama), a statistical software program for Windows^{TR}, and the level of significance was set at $p < 0.05$, two-tailed.

RESULTS

The FIM^{SM} motor, cognitive and total scores ranged from 27 to 91 (median 39), 11 to 35 (median 32) and 44 to 126 (median 71), respectively. The FIM^{SM} motor score correlated negatively (rho = -0.486, $p < 0.05$) and FIM^{SM} cognitive score positively with age (rho = 0.391, $p < 0.05$).

Fig. 1 illustrates the percentages of patients requiring assistance (FIM^{SM} score <5), supervision (FIM^{SM} score = 5) and who were independent (FIM^{SM} score >6) for each FIM^{SM} item. The percentage requiring assistance was the highest for stair item, followed in order by chair transfer, toileting, tub transfer, toilet transfer, dressing lower body, bladder control, dressing upper body, bathing, grooming, bowel control, locomotion, and eating. Table I demonstrates the relationship between each item score and age. Grooming, dressing upper body and bathing correlated negatively with age, while eating,

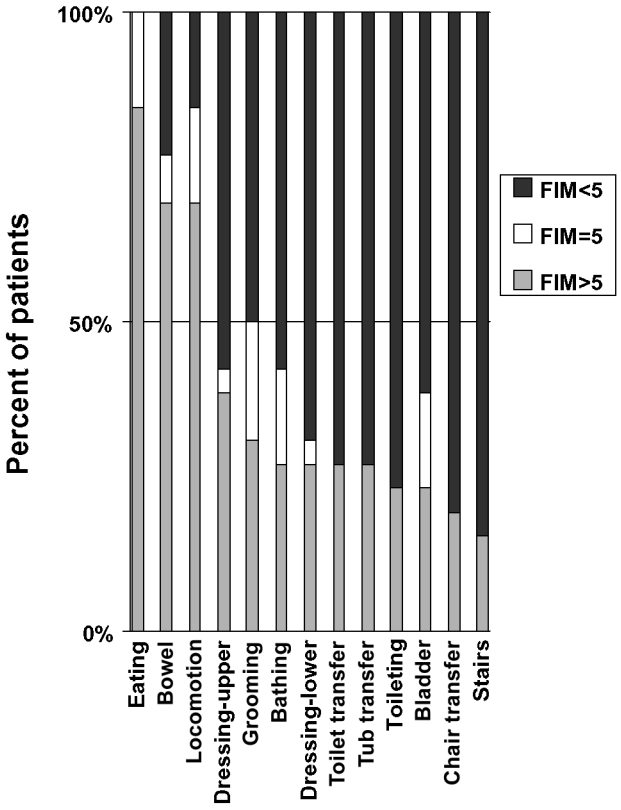


Fig. 1. Percentages of patients requiring assistance (Functional Independence Measure (FIM^{SM}) score <5), supervision (FIM^{SM} score = 5) and who were independent (FIM^{SM} score >5) for each FIM^{SM} item.

Table I. *Correlation between each Functional Independence Measure item and age*

Item	Spearman's rho
Eating	−0.20
Grooming	−0.55*
Bathing	−0.46**
Dressing upper body	−0.50*
Dressing lower body	−0.24
Toileting	−0.31
Bladder management	−0.16
Bowel management	0.34
Bed/chair transfer	−0.34
Toilet transfer	−0.34
Tub/shower transfer	−0.33
Walk	−0.11
Wheelchair	0.47
Stairs	−0.28

*$p < 0.01$, **$p < 0.05$, Spearman's correlation coefficient.

dressing lower body, toileting, bladder and bowel management, chair, toilet, and tub transfers, walk/wheelchair and stair did not.

The average MMT score for the upper limb muscles was 2.75 (SD 0.67, range 1.47–3.86) and that for the lower limb muscles was 2.24 (SD 0.92, range 0.33–3.83). There were significant negative correlations between average MMT scores and age (rho = −0.66 for upper limbs, −0.58 for lower limbs and −0.61 for all 4 limbs, $p < 0.01$).

Mean MMT scores of the upper, lower and all 4 limbs correlated significantly with the FIM^{SM} motor score (rho = 0.65, 0.67 and 0.56, $p < 0.01$) and FIM^{SM} total score (rho = 0.53, 0.55 and 0.77, $p < 0.01$). They did not correlate significantly with the FIM^{SM} cognitive score (rho = −0.19, −0.15 and −0.17, respectively, n.s.).

When we divided the patients into 2 groups according to the FIM^{SM} cognitive score (>25, $n = 21$ and ≤25, $n = 6$), median FIM^{SM} motor score was higher in patients with good cognitive function than those with impaired cognitive function [52.5 (range 37–58) vs 37 (range 30–42), Mann-Whitney U test (14), $p < 0.05$]. The average MMT score, however, was not significantly different between the 2 groups [2.54 (SD 0.80) vs 2.34 (SD 0.73), Mann-Whitney U test, ns] (Fig. 2).

When we analysed the relationship between upper limb muscle strength and the score of the dressing upper body item, patients with average upper limb MMT score ≥3 ($n = 13$) had higher item score than those with average upper limb MMT score <3 ($n = 14$) (median 7 vs 1, Mann-Whitney U test, $p < 0.05$) (Fig. 3a). This held true for the relationship between lower limb muscle strength and the FIM^{SM} bed/chair transfer item score [median 7 ($n = 7$) vs 1 ($n = 20$), Mann-Whitney U test, $p < 0.05$] (Fig. 3b). For locomotion, 11 patients were ambulators

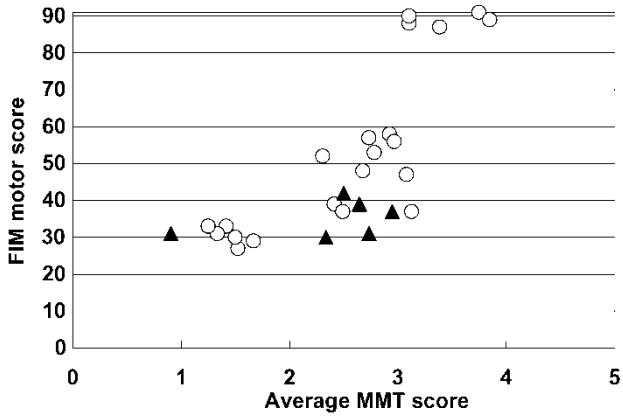
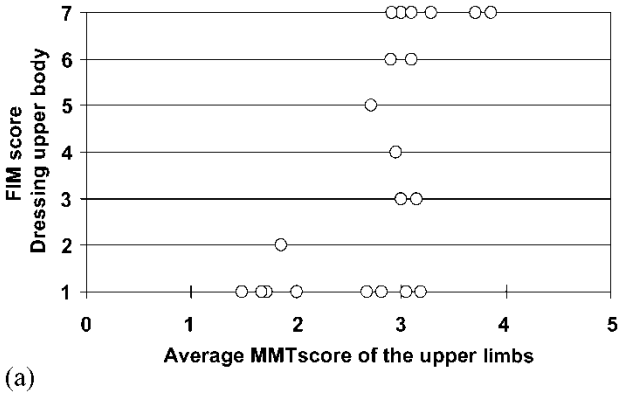
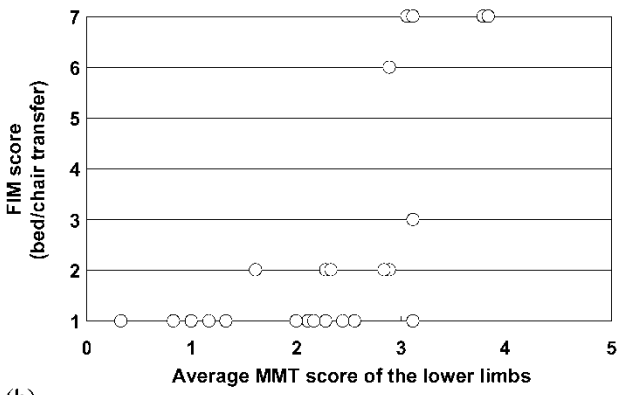


Fig. 2. Relationship between average manual muscle testing (MMT) score of the four limbs and the Functional Independence Measure (FIM^{SM}) motor score ($n = 27$). They were significantly correlated (Spearman's rho = 0.77, $p < 0.01$). When the patients were divided into 2 groups according to the FIM^{SM} cognitive score (>25, $n = 8$ and ≤25, $n = 5$), median FIM^{SM} motor score was significantly higher in patients with good cognitive function than those with impaired cognitive function [52.5 (range 37–58) vs 37 (range 30–42), Mann-Whitney U test, $p < 0.05$], but the average MMT score was not significantly different between the 2 groups [2.54 (SD 0.80) vs 2.34 (SD 0.73), Mann-Whitney U test, n.s.]. ○ = FIM cognitive score >25; filled triangle = FIM cognitive score ≤25.



(a)



(b)

Fig. 3. Relationship between average manual muscle testing (MMT) score of the upper limbs and the Functional Independence Measure (FIM^{SM}) dressing upper body score ($n = 27$) (a) and the score of the lower limbs and FIM^{SM} bed/chair transfer score ($n = 27$) (b). Patients with average upper limb MMT score ≥3 ($n = 13$) had higher item score than those with average upper limb MMT score <3 ($n = 14$) (median 7 vs 1, Man-Whitney U test, $p < 0.05$). Patients with average lower limb MMT score ≥3 ($n = 7$) had higher item score than those with average lower limb MMT score <3 ($n = 20$) (median 7 vs 1, Man-Whitney U test, $p < 0.05$).

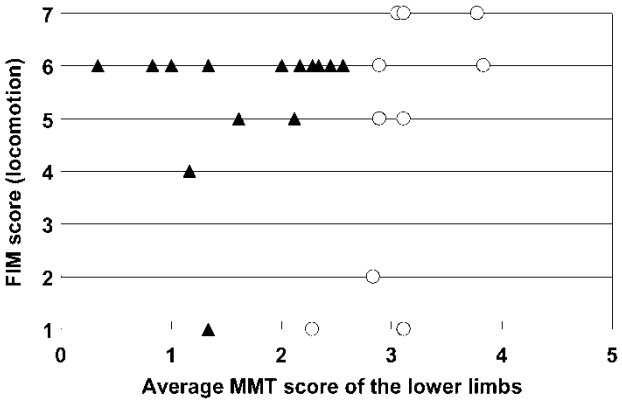


Fig. 4. Relationship between average manual muscle testing (MMT) score of the lower limbs and Functional Independence Measure (FIM^{SM}) locomotion score ($n = 27$). Eleven patients were ambulators and 16 were wheelchair users (12 used a manual wheelchair and 4 a powered wheelchair). None of the patients with average lower limb MMT score <3 were able to walk. ○ = walk; filled triangle = wheelchair.

and 16 were wheelchair users (12 with a manual wheelchair and 4 with a powered wheelchair). None of the patients with average lower limb MMT score <3 were able to walk (Fig. 4).

DISCUSSION

Instrument of activities of daily living

We believe that our study is the first to examine the ADL of patients with DMD in detail using the FIM^{SM}. The FIM^{SM} is a standardized instrument that contains an ADL domain with well-established psychometric properties and is widely used for a variety of disabilities throughout the world (10). Surprisingly, there have been, so far, limited reports studying the ADL of persons with DMD using a standardized instrument. Two studies used the Barthel index (3, 5), and there is 1 study using the FIM^{SM} for children (WeeFIM), a paediatric version of the FIM^{SM} targeted for children aged 6 months to 7 years (21). In the latter report, however, DMD was treated as one of the several disabling conditions of childhood, and detailed information about their ADL is not provided.

The advantages of the FIM^{SM} over the Barthel index are: (i) that it is a 7-point scale instrument, and is potentially more sensitive to detecting deficits in ADL and more responsive to changes than the Barthel index that is a 3-point scale instrument; and (ii) that its scale quality, interrater reliability, construct, concurrent and predictive validity are more fully reported in the literature than are those for the Barthel index (10). Thus, we expected that more detailed and objective information about the ADL of persons with DMD could be obtained with the FIM^{SM}. One can argue that it is more appropriate to use the WeeFIM for this age group, which may be used for children above 7 years of age as long as their functional abilities are below those expected of children aged 7 who do not have disabilities. We selected the FIM^{SM} instead of the WeeFIM because we are interested in following our sample longitudinally and see how their functioning will change as they grow older in our future studies.

Patterns of deficits in activities of daily living

Item difficulties are usually studied with Rasch analysis, or a statistical method to calibrate the scale structure and place the items on a linear scale based on the relative difficulty of the items and the persons' abilities to perform the task items (22). In the present study, we did not perform Rasch analysis because of small sample size. Instead, we tried to infer the difficulty pattern by calculating the percentage of patients requiring assistance (FIM^{SM} score <5). Our results demonstrated that the percentage was the highest for the stair item, followed in order by chair transfer, toileting, tub transfer, toilet transfer, dressing lower body, bladder control, dressing upper body, bathing, grooming, bowel control, locomotion and eating.

Compared with the difficulty pattern reported for Japanese patients with stroke using Rasch analysis (23), the most difficult and the easiest items were similar (stair and eating) (Table II). However, there were number of differences between the 2 populations. In DMD, transfer activities and toileting were among the more difficult items, while toilet and bed transfer items were among the easier items in patients with stroke.

Striking differences were also noted for locomotion and bathing. In DMD, locomotion was the second easiest item following eating, but in stroke, it was the fourth most difficult. This might be explained by the fact that most patients with DMD use wheelchairs (manual or powered) after they lose the ability to ambulate, and remain at the modified independence level at least in the age range studied.

For bathing, it was the second most difficult item for patients with stroke, while it was among the easier items in DMD. This is in contradiction to the study by Lue et al. (3) who found using the Barthel index that bathing was the most difficult item in DMD. To explain this discrepancy, it is worth noting that our patients were younger (mean age 9.7, range 7–14 years) than theirs (mean age 12.4, range 3–24 years). It is also noteworthy that most (85.2%) of our patients were rated either as independent or dependent and only 14.8% were rated as requiring supervision (level 5). When we compared the mean age between the dependent and supervision/independent group, it was significantly higher for the former (10.3 vs 8.7 years, Student *t*-test, $p < 0.05$). This indicates that the transition from independence to maximal or total dependence for bathing takes place very rapidly, and many of Lue's cases might have been older and have already undergone this rapid loss of function. Proof of this speculation awaits future longitudinal studies, but it seems important to take age into account when discussing difficulty patterns of ADL in persons with DMD.

With regard to bladder management, previous studies using the Barthel index reported that they were among the easier items (3, 5). In our study, however, bladder incontinence (FIM^{SM} < 5) was found in 12 of the 27 patients (42.8%). The difference in the scoring between the 2 instruments might explain this discre-

Table II. *Difficulty order of activities of daily living items*

Sample	*n*	Age mean (range) SD	Instrument	Order of difficulty* 1	2	3	4	5	6	7	8	9	10	11	12	13
Stroke (Tsuji et al. (21))	190	61 (14–92)	FIM^{SM}	stair	bathing	tub tr	locomotion	dressing-L	toileting	dressing-U	toilet tr	bed tr	grooming	bladder	bowel	eating
DMD (present study)	27	9.7 (7–14) 2.0	FIM^{SM}	stair	chair tr	bladder	toileting	tub tr	toilet tr	dressing-L	bathing	grooming	dressung-U	locomotion	bowel	eating
DMD (Lue et al. (3))	35	12.4 (3–24)	BI	bathing	stair	toilet tr	ambulation	bed tr	grooming	dressing	bowel	bladder	feeding			
DMD (Nair et al. (5))	32	8.1 (4–13) 2.1	BI	stair	bathing	bed tr	toilet tr	dressing	ambulation	grooming	feeding	bowel	bladder			

*1 = most difficult, 13 = easiest.
DMD = Duchenne muscular dystrophy; FIM^{SM} = Functional Independence Measure; BI = Barthel Index; tr = transfer; dressing-L = dressing lower body; dressing-U = dressing upper body.

pancy. In the Barthel index, they are rated only according to the degree of incontinence, while in the FIM^{SM}, 2 components of management, continence and levels of care, are considered, and the lower score of the 2 is adopted. In our sample, however, all the 12 patients were rated as bladder FIM^{SM} < 5 because of true urinary incontinence. Impaired cognitive function might be another possible reason, but the median FIM^{SM} cognitive score was not significantly different between those with incontinence and those without (32.5 vs 31, Mann-Whitney U test, n.s.). Recently, it has been increasingly recognized that urinary problems are prevalent in patients with DMD, and upper motor dysfunction secondary to spinal deformities and/or post spinal surgery complications is suggested as a possible mechanism (24, 25). Further study is needed to elucidate the reason(s) for their bladder problems.

Factors related to ADL deficits

With respect to factors contributing to ADL deficits, Nair et al. (5) reported that disability as assessed with the Barthel index was related to motor functions as evaluated with motor scores, upper and lower extremity function grades and timed function tests. Lue et al. (3) found that hip contracture and static sitting balance were important factors influencing their ADL.

Our results demonstrated that the motor FIM^{SM} score correlated negatively with age. This indicates that patients with DMD become more dependent in ADL as they grow older, and the loss of muscle strength with ageing is related to this decline in ADL. In fact, we observed significant negative correlations between average MMT scores and motor FIM^{SM} scores and individual FIM^{SM} item scores except bladder and bowel management.

Furthermore, it was impressive that average MMT score of 3 seemed to be an important cut-off point determining the likelihood whether the patient was independent or not both in ADL items requiring upper limb strength (grooming, dressing upper and lower body, and bathing) and those requiring lower limb strength (toileting, bed/chair transfer, toilet and tub transfer, locomotion and stair). When the average MMT score of limb muscles was less than grade 3, persons with DMD tended to need maximal contact or total assistance, but when the average MMT score was equal or above 3, the chances were greater that they were independent in these activities. This did not hold true for eating, whose item score was equal or above 5 regardless of average MMT scores.

It is known that the intelligence quotient of persons with DMD was significantly lower than that for healthy controls (26). Regarding the effects of cognitive function on ADL, our results demonstrated that FIM^{SM} motor score was higher in patients with good cognitive function (FIM^{SM} cognitive score >25) than those with impaired cognitive function, even when their average muscle strength was not significantly different. The reasons for this difference are not clear, but it is important to note that cognitive impairment can adversely affect ADL performance when planning ADL training in DMD.

Limitations of our study

The first limitation is a relatively small sample size. This was because we focused on ADL of people with DMD who were living in the community, in the age range 7–14 years, during which the change in muscular strength and hence in ADL are expected to be large. When we consider the estimated number of people with DMD living in the community in Japan (approximately 2000) (6), we believe that our sample is fairly representative of those living in the community in this age range. To get a more comprehensive view of their disability, however, we need to sample patients in wider age range in the future.

The second limitation might be the validity of averaging MMT scores of major upper and lower extremity muscles, because MMT is not an interval but an ordinal scale. Statistically, this is an area that needs cautious handling, but clinically, MMT is a well-established method of semi-quantifying muscle strength and has been long and widely used in daily practice as well as in clinical trials. Several investigators have used average MMT of representative limb muscles to describe the natural course of the decline in muscle strength (16, 20) and to study the effectiveness of therapeutic interventions (27). Quantitative methods of muscle strength evaluation, such as a simple dynamometer or a more sophisticated isokinetic equipment, might be options to assure linearity of measurement, but the problem of positioning for accurate and reproducible measurement must be considered. We therefore believe that our method of averaging MMT scores is practical and justifiable.

The third limitation would be that we did not examine the contribution of contractures and deformities, which are another important aspect of DMD impairment, to ADL limitations. Lue et al. (3) showed that hip contracture was one of the factors influencing ADL performance in addition to muscle strength.

The fourth limitation might arise from different socio-cultural pattern associated with daily living skills in different societies, for example, dressing, bathing and eating. This factor could limit the generalizability of our findings. In future studies, these aspects of DMD impairment must also be considered.

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