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ORIGINAL REPORT

LIFE HABITS IN MYOTONIC DYSTROPHY TYPE 1

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Objective: **To describe and compare life habits between individuals with adult and mild phenotypes of myotonic dystrophy; identify life habit dimensions in which accomplishment is compromised; and describe satisfaction related to life habits.**

Design: **Cross-sectional study.**

Subjects: **A random sample of 200 subjects with myotonic dystrophy (42 mild phenotypes, 158 adult phenotypes).**

Measurement: **The Assessment of Life Habits (LIFE-H), a questionnaire assessing self-perceived life habits (activities and participation as described in the International Classification of Functioning, Disability and Health (ICF)).**

Results: **Participants with the adult phenotype demonstrated significantly lower participation levels than those with the mild phenotype on 8 out of the 11 categories of the LIFE-H. Lower levels of accomplishment were reported in Mobility, Housing, Fitness, Nutrition, Personal Care, Employment, Recreation, and Community Life categories among the adult phenotype. The Recreation category was the most affected, with 4 out of 7 items revealing compromised accomplishment among 22–27% of individuals. The lowest satisfaction score was observed in the Employment and Recreation categories. In all categories, individuals with the adult phenotype displayed significantly lower satisfaction levels than those with the mild phenotype.**

Conclusion: **This study helped to establish a clearer distinction between activities and participation levels of individuals with the mild phenotype and those with the adult phenotype and supported tailored rehabilitation and community services to improve accomplishment of life habits.**

Key words: activities, participation, myotonic dystrophy, neuromuscular disorders, life habits.

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INTRODUCTION

Myotonic dystrophy type 1 (DM1) is the most common neuromuscular disorder in adulthood (1). It is an autosomal dominant disease with impairments in the muscular, nervous, ocular, digestive, respiratory, cardiovascular, endocrine and reproductive systems (2). The gene defect responsible for DM1 is located on chromosome 19q13.3 and is known as a nucleotide triplet repeat mutation (CTG)n (3). The length of the repetitions is partly correlated with the severity of the disease and the age of onset (4). Several disabilities have been described, including a progressive loss of muscle strength of about 1% per year (5), myotonia, early bilateral cataracts (6), excessive daytime sleepiness (7) and reduced higher cognitive functions (8).

Although considered a highly variable disease with multisystemic involvement, few studies have focused on the impact of DM1 on activity and participation as defined by the International Classification of Functioning, Disability and Health (ICF) (9–11). The latter concept is seen as the result of an interaction process between intrinsic factors including impairments resulting from DM1 and extrinsic factors (Environmental Factors) such as access to healthcare and community services (12). Participation can further be divided into society-perceived and person-perceived participation, which relates to the measurement paradigm (13). The latter is based on an individual's life experiences and preferences, so as to gain a better understanding of an individual's specific needs and problems. A few studies have reported society-perceived participation, mainly regarding the employment dimension in DM1 (14, 15), and person-perceived participation restriction including personal care, housing, mobility and work (9, 11). The present lack of knowledge on this population's experience regarding their perception of their lived experience in carrying out daily activities and social roles hinders the establishment of a long-term management plan for rehabilitation and community services designed for such a population. Indeed, in this population, the planning of cares and services from local agencies have been described as underdeveloped, and medical and paramedical care have been inconsistent, problematic and performed yearly at best (6, 16–18).

Among several issues, the various clinical phenotypes present in DM1 should also be taken into consideration upon establishing a portrait of participation. Classification of DM1 into 4 phenotypes is based on age of onset in conjunction with [CTG]n expansion size: congenital, childhood, adult (classic) and mild phenotype (19, 20). The congenital DM1 (> 1000 CTG) mainly consists of hypotonia at birth, with poor sucking and swallowing, mental retardation, and progressive muscular dystrophy and or-

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gan involvement later on (21). The main features of the childhood DM1 (< 1000 CTG) are onset between 1 and 10 years of age, mild facial weakness, indistinct speech, myotonia and learning difficulties (21). The first 2 phenotypes have a distinct and more severe clinical picture than the latter 2 and will not be discussed in this paper. Individuals with the adult phenotype (100–1000 CTG) demonstrate high symptom heterogeneity, but progressive loss of muscle strength and myotonia are always present. This is the most prevalent phenotype among the DM1 population. Older age of onset (> 40 years old) and minimal involvements such as early cataract and mild myotonia are hallmarks of the mild phenotype (50–150 CTG). It is important to understand that phenotypes are a classification of a continuous spectrum of the severity of the disease as shown by the strong correlations between muscular impairment, CTG repeat number and disease duration (5). Clinically, patients with the mild and adult phenotypes exhibit clearly different pictures and require distinct types of rehabilitation and community follow-ups. However, to our knowledge, no studies have described the differences between their respective levels of activity and participation.

Satisfaction related to participation is increasingly gaining attention in the literature, as it has been associated more strongly with subjective quality of life than the performance component (22). The individual's feelings about or appraisals of his/her participation have thus been suggested as a promising approach in quality of life assessment as well as in healthcare and community services planning and delivery (23). Tailoring our intervention towards the areas demonstrating less satisfaction may improve quality of life more than focusing solely on traditional rehabilitation areas, such as activities of daily living, which only predict a small proportion of quality of life among a neuromuscular population (9).

The objectives of the present study are: (*i*) to describe and compare activity and participation between individuals with the adult and mild phenotypes of DM1; (*ii*) to identify activity and participation dimensions in which the accomplishment is compromised or human assistance is required for a significant sub-sample; and (*iii*) to describe satisfaction associated with activity and participation.

METHODS

Participants

A sample of 200 subjects with DM1 was drawn randomly from a subset of the 416 DM1 individuals listed at the neuromuscular clinic of the Centre de Santé et de Services Sociaux de Jonquière (Quebec, Canada). Participants needed to be over 18 years old, with a diagnosis of DM1 (adult or mild phenotype) confirmed by DNA analysis and able to provide informed consent. For the purpose of this study, subjects were classified as having the mild form of the disease if they presented at least 2 of the 3 following criteria: (*i*) CTG < 200; (*ii*) Muscular Impairment Rating Scale (MIRS) score of 1 (no muscular impairment) or 2 (minimal signs) reported in the medical chart; (*iii*) age at onset of symptoms > 40 years. All subjects who did not fall into this category were classified as having the adult form of DM1. Individuals with other forms of muscular dystrophy, including congenital and childhood DM1 or severe impairments secondary to another disease, were excluded. This study was part of a larger study, which implied 5 days of clinical assessment performed either at the neuromuscular clinic or the participants' homes. From the 416 DM1 individuals listed at the neuromuscular clinic, 82 affected subjects were excluded from the study on the basis of having moved out (36.6%), being unreachable (20.7%), refusing clinical follow-up (20.7%), presenting other major health problems (e.g. tumour) (17.1%), being acquainted with a research team member (2.4%), or other reasons (2.4%); and, 131 other DM1 subjects refused to participate in the study. Reasons for refusal were: lack of interest (58.8%), difficulty moving (12.2%), employment (9.9%), health issues (6.9%), lack of time (4.6%) and various other reasons (e.g. insurance considerations) (7.9%). Finally, 3 subjects dropped out of the study. The institution's ethics committee approved the study.

Instruments

LIFE- H. The short version of the Assessment of Life Habits (LIFE-H) (version 3.1), a 77- item questionnaire was administered at participants' homes by a registered occupational therapist (24). The LIFE-H was based on the Disability Creation Process conceptual framework and designed to be a measure of person-perceived participation. The term "life habits" refers to "daily activities and social roles that ensure the survival and development of a person in society throughout his or her life" (12). Developed prior to the ICF, the taxonomy of life habits covers essentially the dimensions "activities/participation" as presented in the ICF, even though their labels vary slightly: nutrition, fitness, personal care, communication, housing, mobility, responsibility, interpersonal relationships, community life, education, employment, and recreation. In accordance with ICF activity and participation construct, the assessment is based on 2 concepts: the degree of difficulty when performing a life habit in the current environment of the person (no difficulty, with difficulty, accomplished by a proxy, not accomplished or not applicable) and the type of assistance required (no assistance, assistive device, adaptation and/or human assistance). A 10-grade scale of accomplishment was developed by combining the 2 concepts. A grade of 0 indicates the presence of a complete restriction of activity or participation, while a grade of 9 indicates optimal activity or participation. If a specific life habit is not part of one's lifestyle because of personal choice, this item should be marked as not applicable based on the rationale that a restriction of participation cannot be attributed to a life habit that is voluntarily not accomplished. A score for each category can be calculated for each domain (daily activities and social roles sub-score) and for the LIFE-H as a whole (12 categories) ranging from 0 to 10. A higher score indicates a higher level of activity or participation. The Education category is not included as only few participants were expected to be involved in school activities at the time of the study. This instrument has shown adequate test-retest and inter-rater reliability for several populations including DM1 (25, 26).

Muscular Impairment Rating Scale. Participants' muscular involvement was categorized based on the MIRS (27). MIRS is a 5-point ordinal scale used to assess muscular impairment progression in DM1 based on clinical muscular evaluation: 1, no muscular impairment; 2, minimal signs; 3, distal weakness; 4, mild to moderate proximal weakness; and 5 severe proximal weakness.

Statistical analysis

Characteristics of participants are presented with the mean, standard deviation (SD) and range for continuous variables, and frequency and percentage for nominal and categorical variables. As LIFE-H is an ordinal scale, accomplishment and satisfaction scores are presented with the median and the range. The mean and SD are also presented to permit comparison with previous published data. Comparison of characteristics between the mild and adult phenotype was performed with the non-parametric Mann-Whitney *U* test or the χ^2 test. Pearson's correlation coefficient was used to ascertain the relationship between the category accomplishment scale score and its satisfaction score. For objective 2, criteria were established to determine disruption of participation for each life habit items. A grade of 5 or less on the accomplishment scale was chosen as the indicator of a significant disruption because these scores indicate that the life habit is accomplished with human

assistance or not carried out at all. The second criterion was related to the number of individuals who reported significant disruption. A life habit was considered as significantly disrupted when at least 10% of the participants indicated an accomplishment scale grade of 5 or less. The person's level of satisfaction related to the accomplishment of each life habit was also assessed on a 5-point scale ranging from "1 very dissatisfied" to "5 very satisfied". A score of less than 2 on the satisfaction scale was chosen as criterion of significant dissatisfaction. The second criterion was related to the number of individuals who stated significant dissatisfaction. A life habit was considered dissatisfying when at least 10% of the individuals indicated a significant dissatisfaction (scale score less than 2). Statistical analyses were performed using the SPSS statistical software package (28).

RESULTS

Characteristics of participants

The 200 DM1 subjects who completed the study are comparable to the 216 DM1 who did not participate in terms of gender, CTG repeat number and proportion of mild vs adult phenotype but differ slightly in terms of age (mean 47.0 (SD 11.8) years, range 20–81 years, vs 50.2 (SD 14.6) years, $p < 0.05$). However, the age difference is small and should have no impact on the interpretation of the results.

Characteristics of participants for the total group as well as the adult and mild phenotypes sub-sample are presented in Table I. Gender distribution is identical in both groups. For the mild phenotype, more than 50% of our sub-sample showed no or mild muscular involvement, as expected. For the adult phenotype, 53.8% demonstrated mild to moderate proximal weakness in addition to distal muscle weakness. The disease duration for the total group was 21.8 years (SD 9.7).

Table I. *Participants characteristics for the total group and comparison of the mild and adult phenotype sub-samples*

	Total group ($n = 200$)	Adult phenotype ($n = 158$)	Mild phenotype ($n = 42$)
Age, mean (SD) (years)	47.0 (11.8)	44.3 (9.2)	57.4 (14.4)*
Range	20–81		
Gender, *n* (%)			
Men	79 (39.5)	62 (39.2)	17 (40.5)
Women	121 (60.5)	96 (60.8)	25 (59.5)
CTG, mean (SD)	809 (529)	981 (452)	162 (180)*
Range	50–2200		
MIRS *n* (%)			
Grade 1	10 (4.5)	0 (0.0)	10 (23.8)
Grade 2	31 (15.5)	18 (11.4)	13 (31.0)
Grade 3	36 (18.0)	30 (19.0)	6 (14.3)*
Grade 4	98 (49.0)	85 (53.8)	13 (31.0)
Grade 5	25 (13.0)	25 (15.8)	0 (0.0)
Family income (Canadian dollars), % (*n*)	33 (16.5)	30 (19.0)	3 (7.1)
< 10,000	70 (35.0)	66 (41.8)	4 (9.5)
10,000–19,999	38 (19.0)	21 (13.3)	17 (40.5)*
20,000–39,999	20 (10.0)	13 (8.2)	7 (16.7)
40,000–59,999	21 (10.5)	11 (7.0)	10 (23.8)
> 60,000	18 (9.0)	17 (10.8)	1 (2.4)
Unknown/refused			
Education, % (*n*)			
< High school	109 (54.5)	88 (55.7)	21 (50.0)
High school	60 (30.0)	48 (30.4)	12 (28.6)*
College	27 (13.5)	19 (12.0)	8 (19.0)
University	4 (2.0)	3 (1.9)	1 (2.4)

*$p < 0.001$.
MIRS: Muscular Impairment Rating Scale; CTG: cytidine-thymidine-guanidine; SD: standard deviation.

Life habits for the mild and adult phenotypes

Results for each category, domains and total score of the LIFE-H are presented in Table II for the adult and mild phenotypes. In the Daily Activities domain, the Mobility and Housing categories showed the lowest median and mean. In the Social Roles domain, the Employment category was the most disturbed, whereas the accomplishment of life habits in the Responsibility and Interpersonal Relationships categories were preserved among most participants. Participants with the adult phenotype demonstrated significantly lower levels of activity and participation than those with the mild phenotype on 8 out of the 11 (73%) LIFE-H categories. The difference was more than 1 point on the calculated mean score for the Mobility, Housing,

Table II. *Daily activities and social roles domain and categories median and mean scores and comparison between the adult and mild phenotype sub-sample*

	Accomplishment score (/10)			
	Adult phenotype		Mild phenotype	
Category and domain	Median (range)	Mean[a] (SD)	Median (range)	Mean[a] (SD)
Mobility	7.7 (1.9–10)	7.4 (2.3)	9.7 (3.1–10)	8.8* (1.8)
Housing	7.5 (2.9–10)	7.6 (1.8)	9.4 (5.7–10)	8.7* (1.4)
Fitness	9.2 (3.6–10)	8.3 (1.7)	9.7 (6.1–10)	9.3* (1.1)
Nutrition	9.7 (3.6–10)	9.1 (1.9)	10.0 (6.4–10)	9.7* (0.8)
Personal Care	9.8 (4.2–10)	9.2 (1.2)	10.0 (5.8–10)	9.7* (0.7)
Communication	9.8 (7.1–10)	9.6 (0.7)	9.7 (7.1–10)	9.6 (0.5)
Daily Activities sub-score	8.8 (5.0–10)	8.4 (1.5)	9.7 (7.0–10)	9.3* (0.8)
Employment	3.3 (0–10)	4.4 (4.2)	10.0 (0–10)	8.1* (3.6)
Recreation	7.8 (0–10)	6.7 (3.5)	10.0 (0–10)	8.8* (2.4)
Community Life	9.7 (0–10)	8.7 (1.9)	10.0 (5.2–10)	9.5* (1.4)
Interpersonal relationships	10.0 (3.6–10)	9.3 (1.1)	10.0 (4.4–10)	9.4 (1.2)
Responsibility	10.0 (4.6–10)	9.5 (1.0)	10.0 (6.9–10)	9.9 (0.6)
Social Roles sub-score	8.8 (3.9–10)	8.4 (1.5)	9.9 (5.3–10)	9.4* (1.2)
LIFE-H total score	8.7 (4.5–10)	8.5 (1.2)	9.8 (6.2–10)	9.5* (0.9)

[a]Mean score provided for comparison with previous published study.
*$p < 0.001$ (Mann-Whitney *U*-test).
SD: standard deviation.

Fitness, Employment and, the Recreation categories. A similar analysis was performed for each LIFE-H item and significant difference was found for the accomplishment of 37 (48%) life habits, for which individuals with the adult phenotype reported lower performance than those with the mild phenotype. Fig. 1 illustrates the distribution of the most disturbed categories, the domains and the LIFE-H total score. This figure emphasizes that not only the median accomplishment score is lower among participants with the adult phenotype, but also that variability in accomplishment is much higher.

Life habits and gender

Daily Activities and Social Roles sub-scores and LIFE-H total score did not reveal any significant differences between genders. No differences were observed in category scores except the Communication and Employment categories, where women reported higher participation levels.

More severe restriction of life habits and need for human help in DM1

Several life habits items required human help or were not carried out by the participants (37/77; 48%). Fifteen life habits items (41%) in the Daily Activities domain and 22 life habits items (55%) in the Social Roles domain were restricted in some way for over 10% of the sample. The LIFE-H items for which over 20% of the sample reported being unable to accomplish the life habit or needing human help (grade between 0 and 5) are presented in Table III. "Doing major household tasks" and "holding a paid job" were the items most significantly disturbed, with 63.5% and 44.5% of individuals reporting needing human assistance or not accomplishing these life habits, respectively. However, the adult phenotype accounted for most of the participants reporting the need for human help. In the case of the life habit item "doing major household tasks", it could relates to the previous description of miserable living conditions of some individuals with myotonic dystrophy. The Recreation category was the most affected with 4 items (participating in cultural, sporting, outdoor or tourist activities) demonstrating between 23.4% and 35.4% of individuals with a grade below 5.

Table III. *Life habits where participants reported severely disrupted participation (score less than 5)*

Life habits item	% with disturbed participation			*p*-value[a]
	Total	Adult	Mild	
Doing major household tasks	63.5	67.7	47.6	< 0.001
Holding a paid job	44.5	51.9	16.7	< 0.001
Maintaining your home	43.5	50.0	19.0	< 0.001
Maintaining your home yards	42.0	44.0	33.0	< 0.001
Riding a bicycle	33.5	39.2	11.9	< 0.001
Participating in physical activities to maintain fitness	31.5	37.3	1.0	< 0.001
Preparing your meals	27.5	31.2	12.0	0.02
Participating in tourist activities	26.5	30.3	11.9	< 0.001
Getting around on slippery or uneven surfaces	24.0	27.2	11.9	0.04
Taking part in outdoor activities	22.5	39.2	11.9	< 0.001
Getting to public buildings	21.5	24.7	9.5	0.04
Participating in artistic, cultural or craft activities	21.0	24.7	7.1	0.00
Participating in sporting or recreational activities	22.0	24.7	11.9	0.05
Getting to commercial establishments	20.5	24.0	7.1	0.02
Having a sexual relationship	20.5	18.9	26.2	ns

[a]Results of χ^2 test between adult and mild phenotype.
ns: not significant.

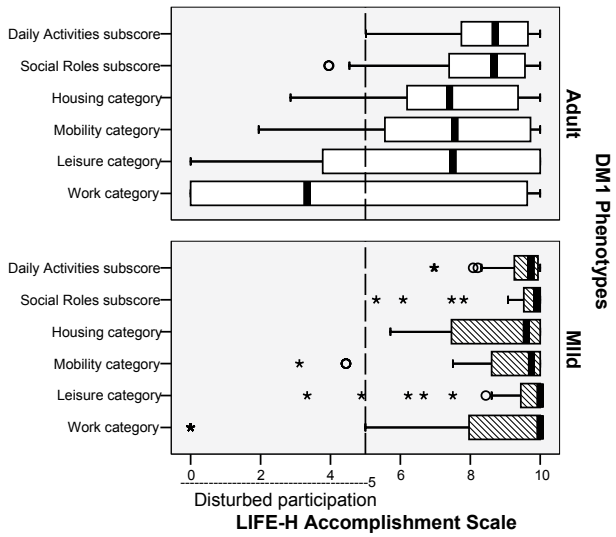

Fig 1. Median, upper and lower quartiles and outliers for the accomplishment score for the most disturbed categories and sub-scores according to phenotypes.
O: cases with values between 1.5 and 3 box lengths from the upper or lower edge of the box.
*: cases with values more than 3 box lengths from the upper or lower edge of the box.
DM1: myotonic dystrophy type 1.

Life habits and related satisfaction

Categories, domains and LIFE-H total satisfaction scores are listed in Table IV. A high level of satisfaction was present for the total and sub-scores for both groups where most participants were satisfied with their participation. The Employment and Recreation categories had the lowest satisfaction scores. Individuals with the adult phenotype reported significantly lower levels of satisfaction for all categories, sub-scores and LIFE-H total score than those with the mild phenotype. The relationships between satisfaction and the accomplishment mean score of the LIFE-H showed significant correlations (r = 0.40–0.84) for all categories, sub-scores and LIFE-H total score. The highest correlations were found for Recreation (r = 0.84) and Employment (r = 0.82) categories. Correlations with satisfaction rating were lower in the Communication (r = 0.40) and Responsibility (r = 0.41) categories, probably

Table IV. *Daily activities and social roles domain and categories satisfaction median and mean scores and comparison between the adult and mild phenotype sub-sample*

	Satisfaction score (/5)				
	Adult phenotype		Mild phenotype		
Category and domain	Median (range)	Mean* (SD)	Median (range)	Mean* (SD)	*p*-value†
Mobility	4.0 (1.5–5.0)	3.9 (0.8)	5.0 (1.0–5)	4.5 (0.9)	< 0.001
Fitness	4.0 (1.7–5)	3.9 (0.7)	4.5 (3.0–5)	4.5 (0.6)	< 0.001
Housing	4.2 (2.3–5)	4.2 (0.6)	5.0 (3.4–5)	4.6 (0.6)	< 0.001
Personal Care	4.6 (2.4–5)	4.4 (0.5)	5.0 (3.8–5)	4.8 (0.4)	< 0.001
Nutrition	4.5 (3.0–5)	4.5 (0.5)	5.0 (3.5–5)	4.8 (0.4)	< 0.001
Communication	4.6 (3.1–5)	4.5 (0.5)	5.0 (3.4–5)	4.8 (0.4)	< 0.001
Daily Activities Sub-score	4.3 (3.0–5)	4.3 (0.5)	4.9 (3.5–5.0)	4.7 (0.4)	< 0.001
Employment	3.5 (1.0–5)	3.3 (1.2)	5.0 (1.0–5)	4.4 (1.0)	< 0.001
Recreation	4.0 (1.0–5)	3.8 (1.0)	5.0 (2.0–5)	4.5 (0.7)	< 0.001
Community Life	4.1 (2.6–5)	4.3 (0.6)	5.0 (2.7–5.0)	4.7 (0.6)	< 0.001
Interpersonal Relationships	4.4 (2.3–5)	4.3 (0.6)	4.8 (2.7–5)	4.6 (0.5)	0.02
Responsibilities	4.8 (3.4–5)	4.6 (0.5)	5.0 (4.0–5)	4.8 (0.4)	0.08
Social Roles Sub-score	4.2 (2.8–5)	4.2 (0.5)	4.9 (3.1–5)	4.7 (0.4)	< 0.001
LIFE-H Total score	4.3 (2.9–5)	4.2 (0.5)	4.9 (3.3–5)	4.7 (0.4)	< 0.001

*Mean score provided for comparison with previous published study. † *p*-value associated with Mann-Whitney *U*-test.
SD: standard deviation.

reflecting the uniformity of respondents' ratings of their level of accomplishment in those life habits.

Table V displays the item in each domain presenting the highest percentage of very dissatisfied or dissatisfied individuals (score 1–2) as well as items presenting ratings below 2 in over 20% of the sample. "Holding a paid job" (Employment category) is the item bearing the highest dissatisfaction level among participants.

DISCUSSION

In any chronic degenerative diseases, the main objective of rehabilitation is to maintain an optimal level of accomplishment in the daily activities and social roles usually valued by the person. Participation has seldom been described in individuals with DM1, making it more difficult to plan adequate rehabilitation and community services. In addition, due to recent advances in molecular diagnosis, individuals with the mild phenotype can be detected much earlier, creating a "new" patient population with specific needs, which has seldom been described to date. Indeed, in the past, only the adult phenotype could have been diagnosed with certainty and offered appropriate medical and rehabilitation services.

Table V. *Life habits items generating high dissatisfaction levels (score 1–2)*

	% with high dissatisfaction			
Life habits items	Total	Adult	Mild	*p*-value*
Holding a paid job	36.4	40.2	15.8	< 0.001
Riding a bicycle	32.8	37.0	15.3	< 0.001
Participating in physical activities to maintain/improve physical fitness/health	28.7	35.0	5.8	< 0.001
Taking part in outdoor activities	24.2	29.6	6.7	0.07
Participating in artistic, cultural or craft activities	22.8	28.4	3.3	0.02
Taking part in unpaid activities	22.4	25.0	15.4	ns

*Results of χ^2 test between adult and mild phenotype.
ns: not significant.

The first objective of this study was to describe and compare activity and participation of individuals with the adult and mild phenotypes. The high results for the global scores (median: <8.7/10) even for the adult phenotype support the clinical observations that most people with DM1 live at home with some services for most of their lives. However, the participation level for more severely affected DM1 subjects is lower than the one found in an aging population even if the mean age group of our sample was relatively younger (44 years old) (29). For most activity and participation categories, the mild phenotype exhibits a higher constant level of participation and the adult phenotype a more heterogeneous level of accomplishment. The Mobility and Housing categories were the most disrupted categories for the Daily Activities domain for both groups and more than 1 point separated the 2 groups. Many activity and participation categories and items rely on muscular strength for the accomplishment of life habits. As 64% of our sample presented some degree of proximal weakness and important distal weakness, it is not surprising to find that Mobility and Housing categories for the Daily Activities domain as well as Employment and Recreation categories for Social Roles domain are the most disrupted and had the lowest satisfaction. In addition, cognitive factors, such as fatigue and lower executive functions known to be present in several adult DM1 individuals, could have also played a role (30, 31). Similar results were found in other studies (10) as well as in our own previous works (5, 32, 33). Social aspects of disablement in DM1 patients were already characterized in a large study conducted in the Saguenay-Lac St Jean region in 1986 (15, 34). Employment among affected males was 3 times lower than in the general population. More than 20% of

affected males and 50% of affected females had never worked and this was reflected in their overall income: 47% of affected individuals being in the lowest income bracket, compared with 25% of the general population, while 42% were estimated to be living below the poverty line. A large proportion of affected individuals (43.6%) compared with the general population (12.2%) depended on social security (15, 34).

The second objective was to identify specific life habits items deserving special attention in rehabilitation and community follow-up. Approximately 20% of life habits items in Daily Activities and Social Roles domains were severely disrupted in our sample. However, individuals with the mild phenotype demonstrated only a sub-optimal level of participation for some life habits items, such as "maintaining their home yards", "performing major household tasks" or "having sexual relationships". The difficulty experienced or the help needed for those life habits could also be related to the normal aging process as our mild phenotype sub-sample had a mean age of 57 years old. The adult phenotype demonstrated more important disruption in several life habit accomplishments such as "taking part in outdoor activities" and "going to public buildings".

The accomplishment level of a life habit is often considered as the main outcome for rehabilitation success. However, as shown in a recent study, satisfaction associated with the realization of this life habit is more related to quality of life than to participation performance (22). A general high level of satisfaction regarding participation in daily activities and social roles was described by participants. Along with the results on the accomplishment scale, Mobility, Recreation and Employment were the most affected categories in addition to the Fitness category. The adult phenotype demonstrated lower satisfaction levels than the mild phenotype on all categories. In general, these subjects reported a higher satisfaction levels than populations of older adults with physical disabilities (29). Such results may appear paradoxical; a lower satisfaction level was expected considering several participants reported difficulty in performing several life habits. However, in our sample, most people had the disease for more than 20 years and they may have shifted their values and expectations (35), thus affecting their satisfaction level. The Housing category provides an example of this situation as several individuals rely on the help of others for several life habits items in this category, although only a small percentage were dissatisfied with their performance. Personality traits, cognitive functions, adaptive capacities and social environment, such as the availability of community services, are factors that should be more closely investigated as possible explanation. In this population, individuals with DM1 are eligible for receiving cleaning services from the community agency to promote independent living. They may be willing to give up such tasks without dissatisfaction, so as to maintain more meaningful life habits. On the other hand, life habits that cannot be carried out with human or technical help or that are usually not part of the healthcare and community services package, such as "riding a bicycle or participating in physical activities to maintain health", tend to induce higher levels of dissatisfaction. In addition, the high dissatisfaction towards participation in unpaid work illustrates a possible avenue for expanding community services, allowing individuals who are unable to work in regular jobs to participate part in important activities, which could be rewarding for them.

These results are important for developing managed care in conjunction with a professional evaluation, since they reveal disrupted participation domains as well as most problematic life habits. The main findings for the elaboration of a care pathway are that: (*i*) the adult and mild phenotypes have a distinct clinical portrait of participation in most life domains and should thus have separate care plans; (*ii*) the adult phenotype displays an heterogeneous picture, thus the need for performing comprehensive evaluation since several aspects may be involved; (*iii*) the mild phenotype presents a near optimal participation but specific aspects need consideration; (*iv*) disruption of several social roles indicates that community management should be part of the care plan. Finally, DM1 being a progressive disorder, the role of environmental factors and especially the implementation of community services should not be underscored as these are most probably effective measures for alleviating some of the consequences and burden imposed by the disease.

Limitations

In interpreting the results, one must bear in mind the limitations of the current study. Concerning the sampling procedure, the individuals who refused to participate in this study may have been more severely or less affected by the disease. The study protocol, a full 5 days of evaluation, was often an issue regarding the participation of full-time working less affected individuals. For more severely affected individual, the most frequent reason for non-participation is the lack of interest, which is a well-known feature of DM1. The lower representation of this population may give an impression that the accomplishment of life habits is higher than expected. However, the same distribution of CTG expansions and the same proportion of mild vs adult phenotype between participants and non-participants suggest a similar disease severity level in both groups. For the measurement aspect, the LIFE-H is a subjective assessment where individuals use their own internal standards to evaluate their accomplishment and is thus subjective to response biases, especially when taking into consideration cognitive deficits, which may be present in DM1 (6). Also, the criterion for disruption of the accomplishment of a life habit was based on the need for human help, but it does not take into account the proportion of participants who reported difficulty achieving specific daily activities or social roles. This may give a more positive evaluation of the problematic as many individuals may experience difficulty, which may progress to the need for external help.

In conclusion, DM1 may be compared to a model of an aging population and this study further supports this hypothesis since, even in their forties, individuals with the adult phenotype may show important disruption in carrying out daily activities and social roles comparable to an older adult population. Tailoring rehabilitation programs and community services more closely to the needs of these individuals will enable better maintenance of life habits and thus will promote increased quality of life.

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REFERENCES

1. Emery AE. Population frequencies of inherited neuromuscular diseases – a world survey. Neuromuscul Disord 1991; 1: 19–29.
2. Harper P, editor. Myotonic dystrophy. 3rd edn. London: WB Saunders; 2001.
3. Fu YH, Pizzuti A, Fenwick RG, Jr., King J, Rajnarayan S, Dunne PW, et al. An unstable triplet repeat in a gene related to myotonic muscular dystrophy. Science 1992; 255: 1256–1258.
4. Gennarelli M, Novelli G, Andreasi Bassi F, Martorell L, Cornet M, Menegazzo E, et al. Prediction of myotonic dystrophy clinical severity based on the number of intragenic [CTG]n trinucleotide repeats. Am J Med Genet 1996; 65: 342–347.
5. Mathieu J, Boivin H, Richards CL. Quantitative motor assessment in myotonic dystrophy. Can J Neurol Sci 2003; 30: 129–136.
6. Harper P. Myotonic dystrophy: a multisystemic disorder. In: Harper P, Van Engelen B, Eymard B, Wilcox D, editors. Myotonic dystrophy: present management, future therapy. Oxford: Oxford University Press; 2004, p. 3–13.
7. Laberge L, Begin P, Montplaisir J, Mathieu J. Sleep complaints in patients with myotonic dystrophy. J Sleep Res 2004; 13: 95–100.
8. Meola G, Sansone V, Perani D, Scarone S, Cappa S, Dragoni C, et al. Executive dysfunction and avoidant personality trait in myotonic dystrophy type 1 (DM-1) and in proximal myotonic myopathy (PROMM/DM-2). Neuromuscul Disord 2003; 13: 813–821.
9. Natterlund B, Ahlstrom G. Activities of daily living and quality of life in persons with muscular dystrophy. J Rehabil Med 2001; 33: 206–211.
10. WHO. International Classification of Functioning, Disability and Health: ICF. Geneva: WHO; 2001.
11. Nätterlund B, Ahlström G. Problem-focused coping and satisfaction with activities of daily living in individuals with muscular dystrophy and postpolio syndrome. Scand J Caring Sci 1999; 13: 26–32.
12. Fougeyrollas P, Noreau L, Bergeron H, Cloutier R, Dion SA, St-Michel G. Social consequences of long term impairments and disabilities: conceptual approach and assessment of handicap. Int J Rehabil Res 1998; 21: 127–141.
13. Cardol M, Brandsma JW, de Groot IJ, van den Bos GA, de Haan RJ, de Jong BA. Handicap questionnaires: what do they assess? Disabil Rehabil 1999; 21: 97–105.
14. Fowler WM, Jr., Abresch RT, Koch TR, Brewer ML, Bowden RK, Wanlass RL. Employment profiles in neuromuscular diseases. Am J Phys Med Rehabil 1997; 76: 26–37.
15. Perron M, Veillette S, Mathieu J. Myotonic dystrophy: I. Socioeconomic and residential characteristics of the patients [La dystrophie myotonique: I Caractéristiques socio-économique et résidentielles des malades]. Can J Neurol Sci 1989; 16: 109–113.
16. Hilton-Jones D. Myotonic dystrophy – forgotten aspects of an often neglected condition. Curr Opin Neurol 1997; 10: 399–401.
17. Fowler WM, Jr., Carter GT, Kraft GH. The role of physiatry in the management of neuromuscular disease. Phys Med Rehabil Clin N Am 1998; 9: 1–8, v.
18. Natterlund B, Ahlstrom G. Experience of social support in rehabilitation: a phenomenological study. J Adv Nurs 1999; 30: 1332–1340.
19. Harley HG, Rundle SA, MacMillan JC, Myring J, Brook JD, Crow S, et al. Size of the unstable CTG repeat sequence in relation to phenotype and parental transmission in myotonic dystrophy. Am J Hum Genet 1993; 52: 1164–1174.
20. Brunner HG, Jennekens FGI, Smeets HJM, de Visser M, Wintzen AR. Myotonic Dystrophy (Steinert's disease). In: Emery AE, editor. Diagnostic criteria for neuromuscular disorders. 2nd edn. London: Royal Society of Medicine Press.
21. de Die-Smulders CE. Congenital and childhood-onset myotonic dystrophy. In: Harper P, van Engelen B, Eymard B, Wilcox D, editors. Myotonic dystrophy: present management, future therapy. Oxford: Oxford University Press; 2004.
22. Levasseur M, Desrosiers J, Noreau L. Is social participation associated with quality of life of older adults with physical disabilities? Disabil Rehabil 2004; 26: 1206–1213.
23. Johnston M, Nissim EN, Wood K, Hwang K, Tulsky D. Objective and subjective handicap following spinal cord injury: interrelationships and predictors. J Spinal Cord Med 2002; 25: 11–22.
24. Fougeyrollas P, Noreau L. Life Habits Measure – shortened version (LIFE-H 3.1). Lac St-Charles, Québec, Canada: RIPPH; 2002.
25. Noreau L, Desrosiers J, Robichaud L, Fougeyrollas P, Rochette A, Viscogliosi C. Measuring social participation: reliability of the LIFE-H in older adults with disabilities. Disabil Rehabil 2004; 26: 346–352.
26. Gagnon C, Mathieu J, Noreau L. Measurement of participation in myotonic dystrophy: reliability of the LIFE-H. Neuromuscul Disord 2006; 16: 262–268.
27. Mathieu J, Boivin H, Meunier D, Gaudreault M, Begin P. Assessment of a disease-specific muscular impairment rating scale in myotonic dystrophy. Neurology 2001; 56: 336–340.
28. SPSS advanced and professional statistics 10.0. Chicago: SPSS Incorporated; 2000.
29. Desrosiers J, Noreau L, Rochette A. Social participation of older adults in Québec. Aging Clin Exp Res 2004; 16: 406–412.
30. Laberge L, Gagnon C, Jean S, Mathieu J. Fatigue and daytime sleepiness rating scales in myotonic dystrophy: a study of reliability. J Neurol Neurosurg Psychiatry. 2005;76:1403-1405.
31. Modoni A, Silvestri G, Pomponi MG, Mangiola F, Tonali PA, Marra C. Characterization of the pattern of cognitive impairment in myotonic dystrophy type 1. Arch Neurol 2004; 61: 1943–1947.
32. Laberge L, Begin P, Montplaisir J, Mathieu J. Sleep complaints in patients with myotonic dystrophy. J Sleep Res 2004; 13: 95–100.
33. Laberge L, Gagnon C, Jean S, Mathieu J. Fatigue and daytime sleepiness rating scales in myotonic dystrophy: a study of reliability. J Neurol Neurosurg Psychiatry 2005; 76: 1403–1405.
34. Veillette S, Perron M, Mathieu J. Myotonic dystrophy: II. Marital status, fertility and gene transmission [La dystrophie myotonique: II Nuptialité, fécondité et transmission du gène]. Can J Neurol Sci 1989; 16: 114–118.
35. Sivaraman Nair KP. Life goals: the concept and its relevance to rehabilitation. Clin Rehabil 2003; 17: 192–202.