THE MYOTONIC DYSTROPHY EXPERIENCE: A NORTH AMERICAN CROSS-SECTIONAL STUDY

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ABSTRACT: *Introduction*: Myotonic dystrophy (DM) is a chronic, multisystemic, neurological condition. Patients and caregivers are uniquely suited to identify what symptoms are most important and highlight the unmet needs that are most relevant to DM. *Methods*: We conducted a North American, cross-sectional study of people with DM type-1, congenital DM, and DM type-2 and their family members. We sent patients and caregivers separate surveys to identify and quantitate the issues of greatest importance, examine the differences between groups, and identify the most important challenges experienced by this population. *Results*: 1,180 people with DM and 402 family members/caregivers responded to the surveys. They reported considerable physical and cognitive symptoms, extensive diagnostic delays, and varying clinical phenotypes on the basis of DM type. *Discussion*: Marked disease burden and numerous unmet needs exist in DM. These needs vary based on DM type and highlight the complex clinical phenotypes of these neurological disorders.

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Myotonic dystrophy (DM) is a multisystemic condition known primarily for muscle weakness, muscle atrophy, and myotonia, along with early cataracts, cardiac conduction defects, central nervous system effects including cognitive deficits and fatigue, and higher rates of cancer.[1–4] There are 2 main types of DM, DM1 and DM2, caused by polynucleotide repeat expansions.[5–8] Myotonic dystrophy type 1 can be further subdivided according to age of symptom onset, including congenital DM1 (CDM), which frequently has additional developmental issues and symptom onset at birth.

Patients with DM can present for medical care secondary to a wide variety of symptoms that occur at variable ages.[9] The diagnostic delay for adults with a confirmed genetic diagnosis has been reported as 7 years for DM1 and 14 years for DM2.[10] Although different symptoms may lead a patient to seek medical care, the most common symptoms are not always those that have the greatest impact on daily life.[1,11]

The Christopher Project gathered data directly from individuals and families with DM across North America. The cumulative perspective of patients and family members has the potential to provide novel understanding of DM experiences and unmet needs concerning diagnostic delays, symptoms, healthcare, and other important aspects of daily life.

MATERIALS AND METHODS

A panel of experts including medical providers, researchers, advocacy organization representatives, caregivers, people with DM, unaffected family members of those with DM, and project partners from the Groupe de Recherche Interdisciplinaire sur les Maladies Neuromusculaires, Marigold Foundation, Muscular Dystrophy Association, Muscular Dystrophy Canada, Myotonic Dystrophy Foundation, Stanford School of Medicine, and The University of Rochester Medical Center worked together to develop a patient survey and a follow-up family member/caregiver survey. Topics, questions, and responses were adapted from previous DM tools, registries, and surveys, including the Myotonic Dystrophy Health Index,[12] Association Francaise contre les Myopathies (AFM) DM1 survey,[13] Naarden Myotonic Dystrophy Consensus data set,[14] and the Myotonic Dystrophy Foundation's patient registry. Others items were added on the basis of suggestions from the panel of experts, patients, and caregivers. The surveys were pilot tested by 20 people with DM, and qualitative phone interviews were conducted to collect feedback to optimize the survey's interpretability and readability. Institutional review board approval was received through Advarra (formerly IRB Services) in both the United States and Canada. Because of the nonclinical nature of the study, implied consent was deemed adequate and was obtained through the respondents' direct participation in the study. Paper-based patient surveys were distributed in 2014 through the project partners' databases to a random selection of database enrollees. Recipients were asked to complete the survey on their own or as a parent/guardian on behalf of a minor (under the age of 18) with DM. Adult patient survey respondents with physical and/or cognitive challenges were asked to report the level of assistance they received, if any, in completing the survey. A follow-up family member/caregiver survey inquiring about their role and perspective as a caregiver and/or family member was

Additional supporting information may be found in the online version of this article.

Abbreviations: CDM, congenital myotonic dystrophy type-1; DM, myotonic dystrophy; ECG, electrocardiogram

Key words: caregiver; myotonic dystrophy; neuromuscular disease; patient report; unmet needs

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distributed in 2015 to those patient survey respondents who agreed to be contacted for additional research, requesting them to ask a family member or caregiver who helps them manage their myotonic dystrophy to fill out the survey.

The patient survey posed 156 questions regarding demographics, diagnosis, symptoms, daily activity challenges, healthcare, insurance, treatments and interventions, and access to information and resources. The occurrence and impact of 29 symptoms were investigated. Symptomatic questions covered areas related to muscle, gastrointestinal function, cardiorespiratory function, sleep, fatigue, and cognitive function. For symptoms that they experienced, respondents were asked if the symptom had "no impact," "minor impact," "moderate impact," or "major impact" on their life, and responses were scored from 1 to 4, respectively. When a participant did not experience the symptom, it was scored as zero. Twenty-three daily life challenges across the following 5 categories were assessed: mobility, household activities, communication, cognitive functioning, and social. Respondents rated the level of challenge in performing each activity as "minor," "moderate," "major," or "unable to perform," and responses were scored from 1 to 4, respectively. Activities that were "not a challenge" were scored as zero. The full patient survey is provided in Supporting Information Materials and Methods 1.

The family member/caregiver survey mirrored the patient survey with 97 questions regarding the symptoms and daily life activities of the person for whom they were caring. Additional questions examined family member/caregiver relationships with the participants and their roles in providing support. The full family member/caregiver survey is provided in Supporting Information Materials and Methods 2.

Responses were grouped according to self-identified DM type. An overall DM group combined all respondents, including those that were unsure of their DM type or who did not provide their type, to capture the complete DM experience. Three groups of DM participants were preidentified for subanalysis. Participants were included in the DM1 or DM2 group when they self-identified as having these conditions. A third CDM group included participants who self-identified with CDM, had been reported to have symptoms at birth, and had received their diagnosis within the first 2 years of life.

Diagnostic delay was defined as the time interval between the reported onset of a patient's first symptom and their age at diagnosis. Respondents who had received a diagnosis before experiencing symptoms were not included in diagnostic delay calculations.

Employment and personal income calculations were performed for respondents aged 16–64 years, and statistical comparisons were performed when cohort size was above 20.

Measures of central tendency and frequency counts were used to summarize results for overall respondents and for different subgroups. Pearson's χ^2 was used to identify statistically significant differences among the different types of DM (CDM, DM1, and DM2) on certain categorical variables. Independent sample t tests were used to measure relative symptom impact and relative daily life challenges by sex as well as frequency of access to specific healthcare providers, time to diagnosis, and age differences for DM1 and DM2 groups. Differences in satisfaction with healthcare across the type subgroups as well as the length of diagnostic delay were measured by using analysis of variance with Duncan *post hoc* tests. By using logistic regression with age as a covariate, statistically significant differences were identified between DM1 and DM2 and between DM1 and CDM regarding whether respondents experienced a symptom (prevalence) and whether they reported any level of challenge performing daily activities. Logistic regression was also used to evaluate income brackets between DM1 and DM2, also controlling for age. Analysis of covariance with age as a covariate was used to test for statistically significant differences between DM1 and DM2 and between DM1 and CDM for symptom impact scores and relative challenge of daily life activity scores. Paired-sample t tests were used to compare reported symptom impact and level of activity challenge between DM patient respondents and family member/caregiver respondents. All tests were performed by using an α level of $P < 0.05$ at a 95% confidence interval. Statistical comparison of DM cohorts regarding prevalence of symptoms and impact as well as prevalence of daily life activity challenges and degree of challenge were corrected by using Bonferroni correction for multiple comparisons, whereby, to achieve $P < 0.05$ after correction, the original uncorrected P-values had to be $P < 0.0017$ for symptoms and $P < 0.0022$ for activity challenges.

RESULTS

Surveys were collected from 1,180 patients and 402 family member/caregivers; respondent demographics are presented in Table 1. Respondents were located in the United States and Canada in 49 states and 9 provinces, respectively. The average age of the DM respondents was 45 years, although this significantly differed between the different DM cohorts. Forty-five percent of respondents had DM1 (of which 165 reported having CDM), and 17% had DM2. Because 30% of respondents either did not know their type or did not provide this information, the overall DM group was used to exemplify the total DM experience across all groups. Seventy-one respondents met the screening criteria for CDM, of whom 69 reported receiving assistance from a parent or caregiver to fill out the patient survey. Seventy percent of respondents with DM had genetic confirmation. Twenty-eight percent of respondents aged 16–64 were working a part- or full-time job or seeking employment; however, 52% stated they were "unable to work due to DM," and 21% earned no income. The full demographic information of respondents in each subgroup is presented in Table 1 and Supporting Information Table 1.

The average delay between symptom onset and diagnosis was 5.6 years (range, 0–48). Participants with DM1 had a significantly shorter time to diagnosis than those with DM2 (Table 1). At diagnosis, 26% of participants received referrals to other healthcare providers, 21% received genetic counseling, and 21% reported receiving no assistance (Supp. Info. Table 2). Participants reported the most helpful assistance at diagnosis were referrals to specialists and other healthcare providers and being directed to patient outreach organizations. A lack of resources regarding DM and the emotional impact of receiving the diagnosis itself were the most commonly reported challenges faced at diagnosis. Although 48% of respondents reported that they would attend a support group were one available, only 6% currently attended such a group. There were no differences between the DM1 and DM2 experience at diagnosis other than the delay in diagnosis.

Table 1. Cohort demographics.

Variables	Overall DM	DM1	DM2	CDM
N	1,180	457	200	71[†]
DM cohort, %	100	39	17	6
Women, %	59	60	65	49*
Genetic confirmation, %	70	77	84	83
Age range, y	0–86	2–81	11–81	0–42
Average age, y ± SD	45 ± 17	45 ± 15	55 ± 14**	14 ± 10**
Average age of onset, y ± SD	26 ± 17	27 ± 15	37 ± 16**	0[‡]**
Average age at diagnosis, y ± SD	30 ± 18	31 ± 15	44 ± 17**	0.2 ± 0.5**
Average diagnostic delay, y ± SD	5.6 ± 8.1	5.7 ± 7.1	7.9 ± 9.4*	0.2 ± 0.5**
Average mutation size, # of repeats ± SD	…	475 ± 718	9,937 ± 9,838	1,447 ± 545
Average mutation size range, # of repeats	…	51–10,000	65–56,000	750–3,400
Median mutation size, # of repeats	…	399	11,270	1,318
Education, ages 25+, *n*	1,006	406	189	11
Some high school, %	6	3	3	27
High school or more, %	92	96	96	64
Some college or more, %	69	79	80	9
Associate's degree or more, %	47	54	59	0
Bachelor's degree or more, %	38	45	46	0
Advanced degree or more, %	10	13	11	0
Employment status, ages 16–64, *n*	945	400	138	27
Student, %	6	7	3	22*
Employed full-time, %	14	17	22	4
Employed part-time, %	11	13	8	19
Retired, %	11	10	17*	0
Unemployed by choice, %	8	13	4*	0
Seeking employment, %	3	4	2	7
Unable to work due to DM, %	52	46	46	48
Unable to work due to other reasons, %	7	5	10*	7
Labor force, ages 16+, *n*	1,084	434	193	27
Labor force participation rate, %	26	32	26	30
Personal income, US respondents ages 16-64, *n*	723	348	113	16
None, %	21	24*	14	38
$1,000–$10,000, %	31	28	20	63
$10,001–$25,000, %	25	24	23	0
$25,001–$40,000, %	7	6	14*	0
$40,001+, %	16	17	29*	0

…, Overall DM cohort mutation data not reported due to respondents having different genetic mutations or lacking genetic testing; CDM, congenital myotonic dystrophy type-1; DM, myotonic dystrophy.

[†]The original number of CDM respondents was 165 but 71 met the preset criteria beyond self-report; this refined cohort is the basis for all future references of CDM.

[‡]An age of onset of 0 (birth) was required to meet the refined CDM cohort criteria.

*$P < 0.05$, **$P < 0.0001$, significant differences between DM1 and DM2 or between DM1 and CDM.

The most commonly reported symptoms in the overall DM group were muscle weakness, fatigue, and daytime sleepiness. These symptoms were also the most impactful. A full list of symptoms and impact for each subgroup is presented in Table 2.

The most commonly used medical practitioner was the family doctor or general practitioner, followed by the neurologist, ophthalmologist, and cardiologist. Eighteen percent of respondents were unaware of DM cardiac risks, and 25% were unaware of DM anesthesia risks. Participants identified topics about which they would like more information, including available treatments and medications, clinical trials, exercise, scientific research, and cardiac implications (Supp. Info. Table 3).

There were 19 daily life activities reported as posing a challenge to the majority with DM. Activities causing the most prevalent challenge related to handling objects, going up and down stairs, and standing. These same activities were also rated as the most challenging. A full list of daily life activity difficulties and degree of challenge for each subgroup is presented in Table 3.

Respondents with DM1 reported significantly higher symptom prevalence in daytime sleepiness, dysphagia, ptosis, and hiccups compared with those with DM2 (Table 2). Respondents with DM2 had a higher prevalence of diabetes. Evaluation of difficulties with daily activities revealed that participants with DM1 had a higher prevalence of difficulty swallowing, dressing, speaking, and alertness (Table 3). Forty-six percent of both the DM1 and DM2 groups reported an inability to work because of DM; however, more participants with DM1 had little or no personal income, and significantly more participants with DM2 reported income in the 2 highest personal income brackets (Table 1).

Participants with congenital CDM had a higher prevalence of learning difficulties compared with other participants with DM1, and the impact of these

Table 2. Prevalence and impact of symptoms in people with DM.[†]

	Prevalence, %				Impact Score			
	Overall DM	DM1	DM2	CDM	Overall DM	DM1	DM2	CDM
Muscle weakness (dystrophy)	94	94	95	94	3.2	3.0	3.3	3.3*
Fatigue	89‡‡	93‡	92	83	2.7	2.9‡	2.7	2.2
Daytime sleepiness	87	93	83*	72	2.5	2.8	2.3**	1.7
Balance issues	82‡	79	87	75	2.5	2.3	2.7	2.3**
Myotonia (difficulty relaxing muscle)	82	88‡	81	69*	2.2	2.3	2.1	1.8
Muscle aches, cramps	79	79	83	63	2.3	2.2	2.4	1.6
Muscle pain	74	72	79	54	2.1	2.0	2.3	1.5
Difficulty swallowing (dysphagia)	67	73	56*	62	1.7	1.8	1.4*	1.6
Constipation	67‡	68‡	67	73	1.7‡‡	1.7‡	1.7‡	2.1
Difficulty concentrating	63	64	64	85	1.6	1.5§	1.6	2.8
Drooping eyelids (ptosis)	62	66	53*	48	1.6	1.7	1.3*	1.1
Anxiety	61‡	63	63	55	1.5‡	1.6	1.5	1.4‡
Abdominal pain	59‡	64‡	48	61‡	1.5‡	1.6‡‡	1.1	1.4
Diarrhea	59‡	64‡	49	54	1.5‡	1.7‡‡	1.2	1.2
Depression	56	60	60	25*	1.4	1.5	1.5	0.5
Difficulty falling asleep	56	58	68	30*	1.5	1.6	1.9	0.7
Balding/thinning hair	52§§	54§§	51§	21§	1.5§§	1.5§§	1.4§§	0.6§
Shortness of breath	52	52	54	27‡	1.2	1.2	1.3	0.7
Learning difficulties/challenges	52§	47§	43	94*	1.4§	1.2§	1.1	3.4**
Abnormal heart rhythm	49	51	53	45	1.2	1.2	1.3	0.9
Frequent hiccups	48	55	33*	58	1.0	1.1	0.7*	1.3
Trouble breathing during sleep (apnea)	47§	50	39§	44	1.3§	1.4§	1.1	1.3
Headaches/migraines	44‡‡	45‡	47	20*	1.0‡‡	1.0‡‡	1.0‡	0.3
Dizziness/fainting	40‡‡	45‡	41	7*‡	0.8	0.9	0.9	0.2
Hearing loss	32	28	48	14	0.9	0.8	1.5	0.3
Sexual/intimacy problems	31§§	32§	44§	3	0.9§	0.9§	1.3	0.1
Recurrent lung infections/pneumonia	27	25	26	48	0.7	0.6	0.7	1.3*
Fertility problems	16‡	18‡	19	3	0.6	0.6‡	0.6	0.1
Diabetes	14	9	27*	1	0.4	0.2	0.7*	0.0

CDM, congenital myotonic dystrophy type-1;DM, myotonic dystrophy.

[†]Statistics were controlled for age differences, and a Bonferroni correction for multiple comparisons was applied. Prevalence calculations were based on group sizes of n = 1,180 for overall DM, n = 457 for DM1, n = 200 for DM2, and n = 71 for CDM. Impact score ranged from 0 to 4.

*P < 0.05, **P < 0.0001, significantly different prevalence or impact of a symptom between DM1 and DM2 or between DM1 and CDM.

‡P < 0.05, ‡‡P < 0.0001, significantly higher prevalence or impact of symptoms in females.

§P < 0.05, §§P < 0.0001 significantly higher prevalence or impact of symptoms in males.

learning difficulties of their lives was significantly higher (Table 2). Although the prevalence of most activity challenges did not differ, the degree of challenge in almost all activities was significantly higher in CDM compared with DM1 (Table 3). Respondents with CDM also reported more referrals to specialists at diagnosis, and only 1% said they were provided no assistance at diagnosis compared with 21% of the overall DM group.

The follow-up family member/caregiver survey was completed mostly by spouses and parents living with the respondents with DM; family member/caregivers reported providing "moderate" or "major" assistance on a daily basis (Supp. Info. Table 4). The most common forms of provided support were emotional, household tasks, attending clinic visits, and financial (Supp. Info. Table 4). Caregivers' responses aligned with respondents with DM in 49 of 52 instances regarding symptoms and daily life challenges (Figs. 1, 2). In all cases in which impressions significantly differed, the caregiver asserted that the prevalence of daily activity challenges was higher for driving, planning daily activities, and intimate life (Fig. 2). Caregivers reported that their most common challenges were managing the DM respondent's complex medical condition (including managing physical and cognitive symptoms and interacting with medical professionals), dealing with the physical and emotional burden on themselves as a caregiver, and providing coaching and support to the respondent with DM (Supp. Info. Table 4). Only 7% of caregivers thought the DM respondent's needs were being met. The most commonly reported unmet needs related to managing symptoms and physical problems, the availability of medical expertise, and the lack of treatments.

DISCUSSION

The Christopher Project study adds to prior smaller cross-sectional studies that sought to define the level of DM disease burden from the patient's point of view.[1,11,15,16] This study again provides evidence that symptoms and challenges are not only common and life-altering, but also under-recognized and under-addressed in the clinical setting. There are few large DM studies that statistically compare symptom

Table 3. Prevalence and level of difficulty performing daily activities for people with DM.[†]

	Challenge prevalence, %				Relative degree of challenge			
	Overall DM	DM1	DM2	CDM	Overall DM	DM1	DM2	CDM
Mobility								
Going up and down stairs	84	79	89	87*	2.3	2.0	2.6	2.3**
Standing for any length of time	83	80	88	77	2.1	1.9	2.2	2.0*
Maintaining balance	81	74	88	80	2.0	1.7	2.0	2.0**
Stand up, sit down, bend down	76	70	83	76*	1.7	1.4	2.0	1.7*
Walking outside or inside	75	70	75	80*	1.7	1.5	1.7	2.0**
Driving a car	44	36	44	65*	1.3	1.0	1.0	3.3**
Household activities								
Handling objects, opening jars, knobs	86‡	88	80	83	2.1	2.1	1.7*	2.6*
Housekeeping, cleaning, laundry	73	70	74	77	1.8	1.5	1.8	2.9**
Preparing meals	59	56	59	76*	1.3	1.1	1.1	2.7**
Swallowing, eating, drinking	59	64	46*	58	1.1	1.1	0.8**	1.3
Using cutlery and kitchen utensils	57	56	49	79*	1.2	1.1	0.9	2.2**
Dressing, doing up buttons, zippers	57§	57§	44*	86*	1.2§	1.1§	0.8*	2.5**
Washing, showering, bathing	53	45	51	80*	1.1	0.8	0.9	2.2**
Communication								
Speaking, pronouncing words	63§	64§	45*	90	1.2§§	1.1§§	0.7	2.5**
Writing, holding a pen	54§§	52§§	50	87*	1.1§§	0.9§§	0.9	2.5**
Cognitive functioning								
Alertness, difficulty staying awake	69	78	58*	55	1.3	1.6	1.1*	1.0
Remembering things	65	64	71	63	1.2	1.1	1.2	1.4
Concentrating	61	61	61	82	1.1	1.0	1.1	1.9*
Putting thoughts into words	55	51	59	86	1.0	0.9	1.0*	2.3**
Planning daily activities	44	41	40	73	0.8	0.7	0.7	2.3**
Social								
Romantic, emotional, intimate life	51§§	50§	53	48	1.3§§	1.1§§	1.3	2.2*
Relationships/interactions with people	48§	49	47	61	0.9§	0.9§	0.8	1.4
Disclosure, talking about their DM	41	41	39	58	0.9§	0.9§	0.8	2.2*

CDM, congenital myotonic dystrophy type-1;DM, myotonic dystrophy.

[†]Statistics were controlled for age differences, and a Bonferroni correction for multiple comparisons was applied. Prevalence calculations were based on group sizes of n = 1,180 for overall DM, n = 457 for DM1, n = 200 for DM2, and n = 71 for CDM. Relative degree of challenge was scored from 0 to 4.

*P < 0.05, **P < 0.0001, significantly different prevalence or degree of challenge of a daily activity challenge between DM1 and DM2, or DM1 and CDM.

‡P < 0.05, significantly higher prevalence or degree of challenge in females.

§P < 0.05, §§P < 0.0001, significantly higher prevalence or degree of challenge in males.

prevalence differences among different DM types. Even after controlling for age, participants reported a higher rate of diabetes in DM2, and, although diabetes has previously been described in DM2, it has not been shown to be significantly different compared with in DM1.[17–19] The prevalence and degree of challenge of the top daily life activity challenges also differed by DM type, providing evidence of the varying clinical needs of each DM type. More research is required to explore further and confirm the phenotypic variance between these groups.

Education, employment, and income varied depending on DM type. The DM1 and DM2 cohorts had a level of education comparable to education attainment norms in the 2015 US Census and the 2016 Canadian Census.[20,21] Despite their level of education, respondents had vastly reduced participation in the labor force, which is defined as those aged 16 years and older who are employed part-time or full-time or who are actively looking for work.[22] In the same year that study data were collected, the US labor force participation rate was 63% and the Canadian rate from 1 year later was 65%,[23] whereas the DM group had only 26% participation. Fewer US respondents with DM earned in the highest 2 personal income brackets compared with US norms[24]; the greatest disparity was with CDM, followed by DM1, and then DM2.

The DM diagnostic experience is complex, reaching well beyond delivery of the actual diagnosis, and ideally involves an engaged healthcare provider to address the variety of DM patient needs. Participants had an average 5.6-year diagnostic odyssey and, although it is encouraging that this delay is shorter than that reported in previous studies,[10] this time frame remains long and represents an addressable burden to patients, families, and the healthcare system. Improved communication and education at the time of diagnosis are essential; 21% of respondents with genetic confirmation were still unsure of their DM type. Knowing one's DM type is important because it helps with disease management, helps patients understand their condition better, and has implications for family planning. Many respondents also reported inadequate assistance beyond the time of diagnosis; 71% wanted more information about DM, and 38% had unmet health care needs such as consistency and expertise of medical care.

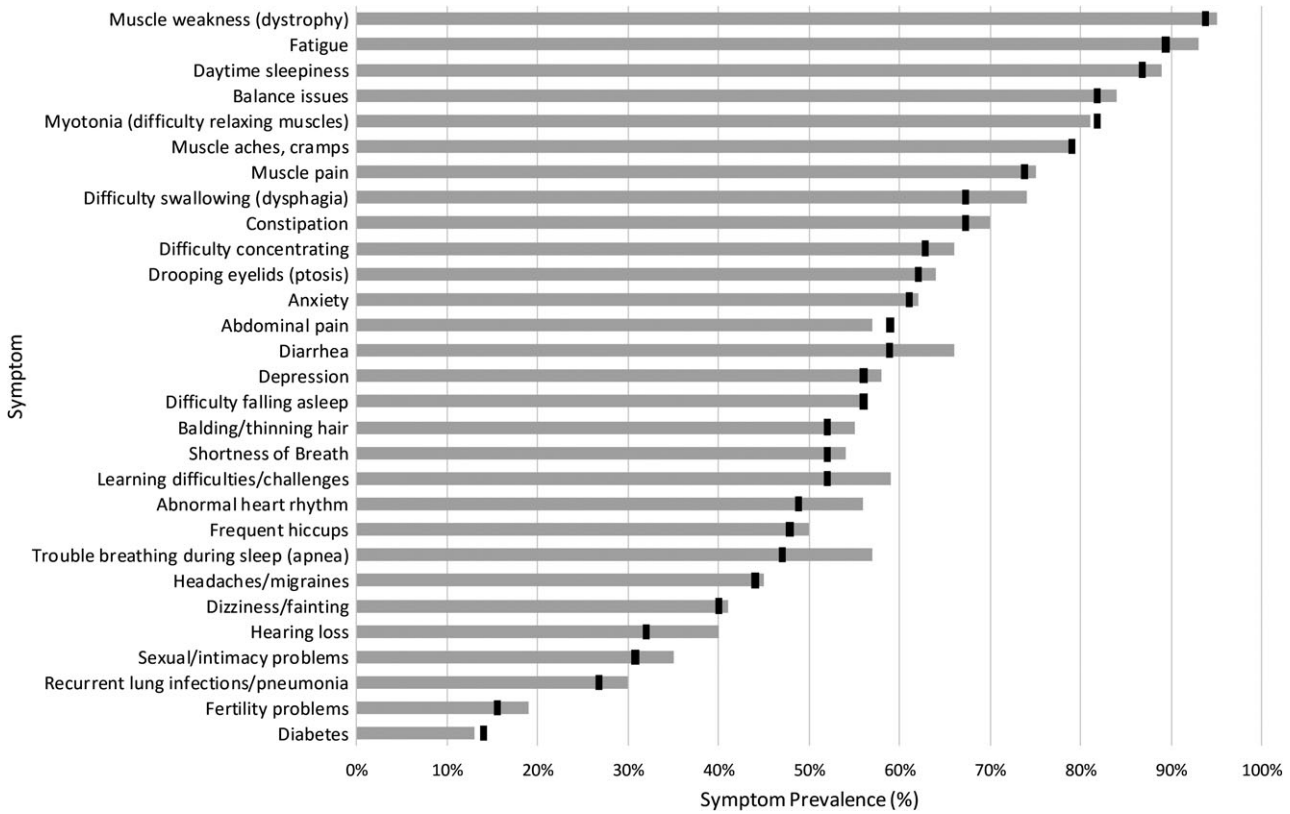

FIGURE 1. Family members' and caregivers' impressions of the myotonic dystrophy respondents' symptom prevalence in comparison to respondents' assessments. Horizontal gray bars represent the family member/caregiver report on patients' symptom prevalence. Vertical black tick marks represent the corresponding patient-reported prevalence value. No significant symptom prevalence differences were found between caregivers and myotonic dystrophy respondents.

Lack of available DM information can have life-threatening consequences. Anesthesia should be carefully planned in patients with DM to prevent problems caused by sedatives and analgesics,[2] yet 25% of respondents with DM were unaware of these risks. Anesthesia complications have been reported in 7%–18% of surgical procedures involving patients with DM.[25,26] Anesthesia risks can be reduced through the careful selection of anesthetics and vigilant monitoring during and after procedures.[27] Cardiac monitoring in the form of annual electrocardiograms (ECG) or Holter monitors is similarly important in DM to identify progressive, potentially life-threatening cardiac arrythmias,[28] yet 18% of respondents with DM were unaware of their cardiac risks. Mortality in DM is sevenfold higher than in the general population,[29] and one study found that 88% of asymptomatic individuals with DM had conduction defects.[30] Another study found that 24% of patients with DM1 had severe ECG abnormalities, and 20% had died within 5 years, with 33% of those deaths being sudden.[31] Because severe ECG abnormalities can predict sudden death and implantable cardioverter defibrillators may prevent some episodes of sudden death in DM, cardiac monitoring and interventions by a trained cardiologist aware of the intricacies of DM is warranted.[31,32]

This study has limitations inherent to a large cross-sectional, self-report survey. Specifically, the sample population may not perfectly represent the DM community. Although recruitment was through a variety of organizations, some DM patients may not be accessible through these mechanisms. Responding individuals may also be more engaged or have more resources available than the general DM population. In addition, more responses came from women than from men, perhaps resulting in a bias toward sex-specific responses. The study was also limited by relying on respondents' accurate memory of historical facts about their diagnostic experience. Respondents also included patients who self-reported having DM without confirmation through medical record review, in contrast to prior studies that required vetting of medical records.[1,11] Regardless, this study again provides evidence that select groups with DM are capable of filling out a self-report on the occurrence and relative importance of their clinical symptoms.[1,11,12,16,33]

One of the key findings of the Christopher Project is that additional information regarding DM is required, a deficiency that this research helps address. In recognition of this requirement, a lay summary of the research will be openly shared with respondents and the

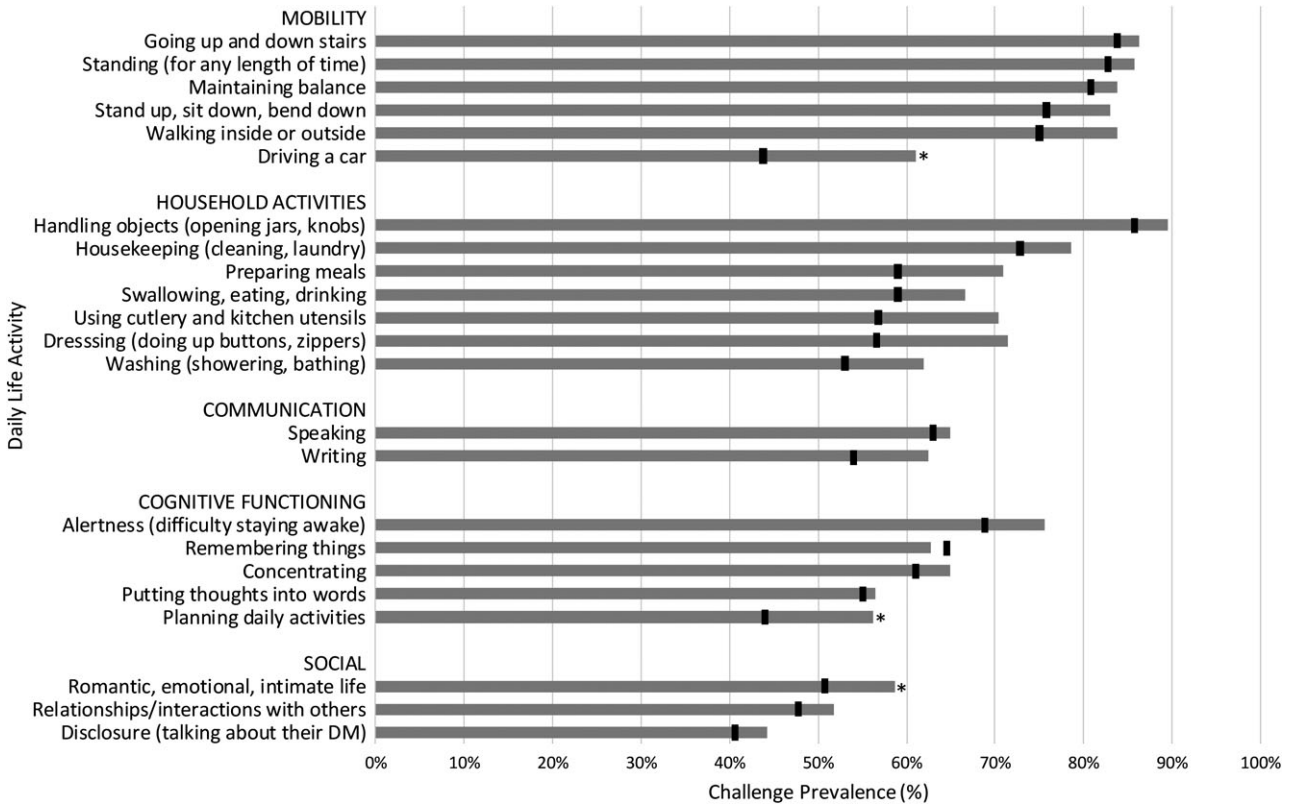

FIGURE 2. Family members' and caregivers' impressions of the myotonic dystrophy respondents' daily life activity challenges in comparison to respondents' assessment. Horizontal gray bars represent the family member/caregiver report on patients' daily life activity challenge prevalence. Vertical black tick marks represent the corresponding patient-reported prevalence value. $^{*}P < 0.05$ comparing patient report to the family member/caregiver report.

DM community.[34] The deidentified raw data will also be accessible to researchers under a formal application and review process.

This research highlights a requirement for better education throughout the patient's disease course, a more efficient diagnosis, and a timelier acknowledgment of the potentially serious risks associated with DM through routine clinical care. Healthcare providers have the opportunity to educate themselves further on these novel diseases, provide information to patients, and link patients to appropriate resources and support so that their patients can make tangible contributions to their own health.[35] Advocacy groups can also develop and disseminate accessible and understandable resources and care guidelines that educate the community about DM to help address unmet medical needs, improve the diagnostic experience, and raise awareness about risks.

Our data show the considerable disease burden and unmet needs of those with DM and their families. Although more research is required, findings from this study have the potential to improve health outcomes for people living with DM. Overall, the Christopher Project provides extensive patient and caregiver insight into a complex, multisystemic, and understudied neurological condition for the benefit of the entire DM community.

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