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Understanding the burden of care in myotonic dystrophy: Findings from a caregiver survey in Canada and the United States

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Abstract

Background:

Myotonic dystrophy (DM) is a progressive, multisystemic disorder affecting muscular, cardiac, respiratory, endocrine, and central nervous systems, with cognitive and behavioural symptoms that create complex, long-term caregiving demands. Although neuromuscular disease caregiving involves substantial emotional, financial, and time-related strain, caregiver experiences in DM remain understudied.

Methods:

The Myotonic Dystrophy Foundation surveyed 292 caregivers in the United States (92%) and Canada (8%) supporting individuals with DM1 (83%) and DM2 (13%). Caregivers answered questions about their relationship to DM care (relationship, number cared for, level and type of care) and their caregiving experience (support, respite breaks, and caregiver burden). Analyses included descriptive statistics, Wilcoxon signed-rank tests, Spearman correlations, and OLS regression using a seven-item caregiver burden index ($\alpha = 0.82$). Thematic analysis was conducted on an open-ended response, and findings were validated through a caregiver focus group.

Results:

Respondents were predominantly female (77%), white (92%), and over age 50 (84%). Over a quarter (28%) provided major care, and 52% moderate care. Pairwise Wilcoxon tests (Bonferroni $\alpha = 0.00238$) showed that mental exhaustion exceeded all other caregiver burdens ($z = 5.57–12.34$, $p < .001$), including financial burden, health impacts, worries, and ability to care for others, while physical exhaustion was lower than mental ($z = -4.38$, $p < .001$) but higher than others ($z = 3.30–11.44$, $p \leq .001$). The seven burden domains were strongly interrelated, reflecting cumulative emotional, physical, and practical demands. Regression indicated caregiver burden is significantly associated with level of care and frequency of breaks, with higher burden for moderate (0.347, $p = 0.024$) and major care (0.567, $p = 0.002$) and for caregivers with one to two breaks a year or no breaks (0.422–0.447, $p \leq 0.01$). Age, number of individuals cared for, and availability of support was not significant. The model explained 15% of variance ($R^2 = 0.146$, F $p = 0.0028$).

Conclusions:

Caregivers experience interconnected emotional, physical, and practical challenges that intensify with greater care needs and limited respite. Findings highlight the importance of accessible, meaningful respite and coordinated support strategies addressing the cumulative nature of caregiver burden in DM.

Background

Myotonic dystrophy (DM) is the most common adult-onset muscular dystrophy, with two genetic subtypes: type 1 (DM1) and type 2 (DM2). Both are multisystemic conditions that lead to progressive muscle weakness and affect respiratory, cardiac, endocrine, ocular, gastrointestinal, and central nervous systems [1–4]. Disease progression is highly variable, with DM1 typically presenting earlier and with more pronounced systemic involvement, while DM2 generally manifests later with proximal weakness and typically more gradual progression [1–4]. The multisystemic nature of DM, which includes both cognitive and physical symptoms, results in complex care needs and progressive disability, often requiring sustained family and extended caregiving across the lifespan.

Caregiving needs associated with DM extend beyond physical assistance alone. Individuals with DM frequently experience cognitive and behaviour changes such as apathy, executive dysfunction, and hypersomnolence, which can complicate disease management [5–10]. A recent review of the Myotonic Dystrophy Family Registry (N = 1,744), which includes participants with DM1 and DM2, found that the most prevalent self-reported symptom by all respondents, regardless of DM type or age, was daytime sleepiness at 76.6% [11]. Furthermore, psychological and behavioural features common in DM, including depression, anxiety, apathy, and impaired coping in affected individuals may increase complexity of care [5, 7]. The demands of long-term care for cognitive, behavioural, and physical disease symptoms can complicate disease management and place significant strain on families, highlighting a potential need for additional support.

In DM and broader neuromuscular disease (NMD) populations, caregiving burden also includes substantial economic impacts, such as increased medical costs, lost income, and reduced workforce participation [12–15]. Caregivers in NMD populations additionally report limited respite opportunities and inadequate information from healthcare professionals, further intensifying long-term care demands [16–17].

In addition to emotional and financial consequences, caregiving in DM has implications for caregivers' physical health. Analyses of comorbidities and healthcare utilization show that families providing prolonged care for individuals with NMD conditions face a higher risk of their own health complications [17]. Prior caregiver studies indicate that relatives of individuals with DM often provide daily support for medical care, symptom management, and activities of daily living, frequently with limited external resources [17]. Importantly, many caregivers are themselves aging parents or spouses, which intensifies vulnerability as they balance aging with increasing responsibilities [16–17]. Together, these findings underscore the complex and cumulative nature of caregiver burden and the need for targeted research on how to best provide support and resources specific to DM.

Although caregiver burden has been examined in related NMD populations, the unique combination of progressive physical disability and cognitive-behavioural involvement in DM may create a distinct caregiving experience that remains poorly understood. Existing DM caregiver research is limited, particularly regarding how different dimensions of burden interact and which factors most strongly

contribute to challenges for caregivers. The Myotonic Dystrophy Foundation (MDF) developed the DM INSIGHT (Investigating Needs, Support, Gaps, and Healthcare Trends in Myotonic Dystrophy) study in part to evaluate caregiver experiences in Canada and the United States. The study aims to characterize the emotional, physical, financial, and logistical dimensions of caregiver burden, identify major caregiving challenges and support gaps, explore factors associated with increased caregiver strain, as well as inform more comprehensive, family-centred care strategies for caregivers of people living with DM.

Methods

The DM INSIGHT study was designed, in part, to examine the experiences of caregivers of individuals with DM. Specifically, it aimed to capture caregivers' perspectives on potential economic, time-related, social, and physical burdens, as well as how they identify and provide the support needed by individuals living with DM.

Survey development

The tailored caregiver survey was developed by a collaborative team of MDF staff members to assess the needs, experiences, and healthcare challenges within the DM community. Survey items were either created by internal experts or adapted from the Christopher Project's Family Survey, or the Rare Disease Diversity Survey, developed via a collaboration between the Rare Disease Diversity Coalition (RDDC) and the National Organization for Rare Disorders (NORD®) to address the specific needs of DM caregivers [18–20]. The survey was estimated to take approximately 20 minutes to complete and was hosted via Qualtrics. Customized survey links were used to route respondents to the appropriate survey version and monitor recruitment source in order to support duplication prevention. No personally identifying information was used in the analytic dataset.

Participant recruitment

The DM INSIGHT Caregiver Survey was piloted in April 2025 by two caregivers with diverse DM caregiving experiences. It was subsequently distributed in person to caregivers of individuals with DM at the MDF's Conference in Indianapolis, Indiana, in May 2025. After the initial distribution, outreach and study recruitment methods included targeted email campaigns via the MDF database, social media posts, website links, flyers, and electronic communications through the Myotonic Dystrophy Family Registry sent from May to July 2025.

Survey respondents provided informed consent prior to completing the survey. Respondents either lived in Canada or the United States and were at least of the age of majority in their province or state. Caregivers needed to have provided care, informal or paid, to at least one person with a DM1 or DM2 diagnosis. All respondents needed to be able to read and understand English.

Caregiver survey

The DM INSIGHT Caregiver Survey was comprised of seven sections. Following completion of a background questionnaire, caregivers were asked a series of questions regarding their relationship to DM. Adapted from the Christopher Project Family Survey, respondents reported the number of individuals with DM for whom they provided care and identified their relationship to each individual, beginning with the person they considered most affected by the disease.

For each individual cared for, starting with the most affected, caregivers indicated both the level and type of assistance provided. Levels of assistance were categorized as minor assistance (support provided occasionally, with the individual usually independent), moderate assistance (support provided frequently or often), and major assistance (support provided constantly or all of the time).

Caregivers then completed a section assessing their caregiving experience. Items were adapted from the Rare Disease Diversity Survey developed by the RDDC and NORD® [20] and assessed seven domains related to DM caregiver burden: physical exhaustion, emotional or mental exhaustion, financial challenges, personal health issues, physical strain, concerns about the future, and difficulty supporting other family members. Respondents rated the extent to which each item described their experience on a 5-point Likert scale ranging from “does not describe me” to “describes me extremely well.”

In addition to the burden assessment, caregivers reported the frequency of respite breaks taken within the past 12 months, with response options of “never”, “once or twice a year”, “once or twice a month”, “once a week”, or “daily (even if only for a short period each day)”. The survey assessed frequency of breaks but not their duration, quality, predictability, or whether they fully relieved caregivers of supervisory responsibility.

The survey also asked caregivers to identify their greatest challenge in supporting individuals with DM and indicated whether they received support from others, including family members or paid care providers.

Subsequent sections assessed caregivers’ awareness of DM-related information, health insurance and financial considerations, and access to quality healthcare for individuals with DM. The survey concluded with an open-ended question inviting respondents to share any additional comments or experiences.

Data analysis

All descriptive and statistical analyses were performed using Stata/SE 15 (StataCorp LLC, College Station, TX, USA). The seven items on caregiver burden were used to create a single caregiver burden index specific to DM. Internal consistency of the scale was high (Cronbach’s $\alpha = 0.82$). A caregiver burden score was calculated for each respondent as the mean of available items, with higher scores indicating greater burden. Missing data were minimal, with 61 missing items.

Characteristics of individuals with DM and caregivers were summarized descriptively. Denominators vary by item due to missing responses and skip patterns. Frequencies and percentages were used to

summarize categorical variables, while means and standard deviations (SDs) were used to summarize continuous variables. Categorical variables were summarized using frequencies and percentages. Continuous variables were summarized using means and standard deviations (SDs) for approximately normally distributed data, or medians and interquartile ranges (IQRs) for non-normally distributed data. For paired comparisons of continuous variables, the Wilcoxon signed-rank test was used for non-parametric data.

Correlations between variables were tested using Pairwise Spearman. Additionally, the burden index was treated as continuous, based on the $\alpha = .82$ reliability, and linear regression (OLS) was used to understand how the caregivers' age, level of care provided, the ability to take breaks, the existence of other available paid or unpaid support, and the number of people under care predicted burden. For the caregiver age category, sparse age categories (1 or less) were excluded, which dropped caregivers aged 18–29 from the regression analysis.

Eighty-six caregivers provided responses to the open-ended question inviting additional comments. Qualitative findings were used to contextualize survey results rather than to generate independently generalizable qualitative conclusions. Responses were analysed using thematic coding. A total of 17 a priori and emergent codes were applied by a primary reviewer and validated by a secondary reviewer for inter-rater reliability. Each response was coded as a single, holistic unit; multiple codes were not assigned to the same statement. In addition to codes capturing administrative questions and requests for additional support from the MDF, thematic categories reflected several substantive domains. These included healthcare experiences (e.g., navigation, care coordination, insurance-related challenges, and healthcare satisfaction or dissatisfaction) and caregiver burden (e.g., physical exhaustion, emotional or mental exhaustion and burnout, social isolation, financial strain, and future uncertainty). Additional themes included communication and information needs, experiences of loss, hopes for a cure or research advancements, and positive aspects of caregiving.

After an initial analysis, findings were shared with a small focus group of six caregivers in Canada and the United States. Caregivers were engaged to assess whether the results cohered with their lived experiences and to provide feedback on the accuracy and appropriateness of our initial interpretations. After the focus group, the interpretation and contextual framing of the findings were refined to reflect caregiver feedback, while the underlying findings themselves remained unchanged.

Results

Sample population

Two-hundred and ninety-two family caregivers to individuals living with DM responded to the survey, including 92% from the United States (255/277) and 8% from Canada (22/277). Survey respondents are predominantly female (77%, 211/275), white (92%, 257/280), and over 50 years of age (84%, 232/275), with 26% over 70 years of age (71/275). A larger percentage of caregivers are caring for individuals with DM1 (83%, 219/263) than DM2 (13%, 35/263). The majority of caregivers that responded are assisting

one person with DM (64%, 167/259). Respondents could select multiple employment types, and over a third of caregivers (37%, 112/306) indicated they are retired from employment, as seen in Table 1.

Table 1
Caregiver demographic characteristics

Age, years, n (%) (N = 275)	
18–21	1 (0%)
22–29	0 (0%)
30–39	13 (5%)
40–49	27 (10%)
50–59	66 (24%)
60–69	95(35%)
70+	71 (26%)
Prefer not to say	2 (1%)
Gender, n (%) (N = 275)	
Female (including transgender women)	211 (77%)
Male (including transgender men)	56 (20%)
Non-binary	1 (0%)
Prefer not to say	3 (1%)
Prefer to self-identify	(2%)
Race/ Ethnicity, n (%) (N = 280)	
American Indian, Native American, First Nations, or Alaska Native	5 (2%)
Asian	3 (1%)
Black or African American	0 (0%)
Native Hawaiian	0 (0%)
Other Pacific Islander	0 (0%)
Latinx	4 (2%)
White	257(92%)
Other	5 (2%)
Prefer not to say	6 (2%)
DM- related characteristics	
Diagnosis of individual with DM, n (%)* (N = 263)	

Age, years, n (%) (N = 275)	
Myotonic Dystrophy Type 1 (DM1)	219 (83%)
Myotonic Dystrophy Type 2 (DM2)	35 (13%)
Unsure	9 (3%)
Number of individuals with DM in care, n (%) (N = 259)	
1 person	167 (64%)
2 people	50 (19%)
3 people	32 (12%)
More than 3	10 (4%)
Relationship to the most affected person, n (%) (N = 268)	
I am their child	7 (3%)
I am their parent/guardian	136 (51%)
I am their sibling (brother or sister)	6 (2%)
I am their spouse/partner	108 (40%)
Other	11 (4%)
*This is based on the diagnosis of the most affected person.	

Caregiver experience

The survey asked caregivers about the level of support they provide to the individual most affected by DM. Over a quarter (28%, 73/264) reported providing major assistance, offering support constantly or all the time. Over half (52%, 138/264) provided moderate assistance, supporting the individual frequently or often. An additional 19% (51/264) provided minor assistance, offering occasional support while the person was generally independent.

Among caregivers who reported providing major assistance to the person they identified as most affected by DM, the most frequently endorsed activities were assisting with healthcare management and attending medical appointments (98%), followed by emotional support (92%), support with personal care needs (e.g., bathing, dressing) (90%), assistance with household tasks and chores (88%), and financial support (74%). Caregivers reporting moderate assistance most commonly indicated their involvement in healthcare management (92%) and attendance at medical visits (91%), as well as assistance with household tasks and chores (90%), provision of emotional support (86%), and financial support (69%). Among those providing minor assistance, emotional support was most frequently reported (88%). These caregivers also indicated assisting with healthcare management and attending medical visits (69% each) and providing financial support (65%).

Assistance with personal care needs was more common among those providing major assistance (90%) and declined substantially among those providing moderate (43%) and minor assistance (12%). In contrast, involvement in healthcare management and attendance at medical appointments was consistently reported across all levels of assistance (69–98%). Helping with schoolwork was the least frequently cited method of assistance across all groups.

The survey assessed seven aspects related to DM caregiver burden, described in Table 2. Pairwise Wilcoxon signed-rank tests (Bonferroni-adjusted $\alpha = 0.00238$, which implies 21 pairwise comparisons) showed that mental or emotional exhaustion was higher than all other domains, including financial burden ($z = 8.55$, $p < .001$), personal health impacts ($z = 10.77$, $p < .001$), strength needed for care ($z = 5.57$, $p < .001$), worries about the future ($z = 6.47$, $p < .001$), and ability to care for others ($z = 12.34$, $p < .001$).

Table 2
Descriptive statistics of caregiver burden index (Scale: 1–5)

Component	Number	Mean	Standard Deviation
Mental or emotional exhaustion	242	3.25	1.14
Physical exhaustion	244	2.95	1.15
Strength needed for physical care	244	2.84	1.24
Worry about the future	243	2.67	1.32
Personal health impacts	242	2.24	1.13
Financial burden	243	2.22	1.34
Ability to care for others in their family	243	1.77	1.03

Physical exhaustion was lower than mental exhaustion ($z = -4.38$, $p < .001$) but higher than all other domains, including financial burden ($z = 7.27$, $p < .001$), personal health impacts ($z = 9.10$, $p < .001$), worries about the future ($z = 3.30$, $p = .001$), and ability to care for others ($z = 11.44$, $p < .001$); it did not differ from strength needed for care ($z = 2.34$, $p = .019$) as this comparison did not meet the Bonferroni-adjusted threshold.

Financial burden and personal health impacts did not differ ($z = -0.63$, $p = .526$) and fell in the mid-range. Financial burden was lower than strength needed for care ($z = -6.36$, $p < .001$) and worries about the future ($z = -4.40$, $p < .001$), but higher than ability to care for others ($z = 4.71$, $p < .001$). Personal health impacts were also lower than strength needed for care ($z = -7.71$, $p < .001$) and worries about the future ($z = -5.31$, $p < .001$). The mid-range positioning of financial burden and personal health impacts, and their lack of difference from one another suggest these are meaningful but not as dominant stressors for the survey respondents.

Strength needed for care did not differ from worries about the future ($z = 1.54$, $p = .124$). Ability to care for others was lower than all other domains. The consistently lowest ratings for ability to care for others may indicate that, despite high levels of exhaustion, caregivers may still perceive themselves as maintaining their responsibilities for caring for other members of their family, potentially at a personal cost of exhaustion and health impacts. However, lower scores on this item may indicate either less perceived impact in this domain or variability in item relevance depending on family structure.

Using pairwise Spearman correlations, we examined relationships among the seven domains. Physical exhaustion showed strong positive correlations with the physical strength needed for care ($\rho = .65$, $p < .001$), mental exhaustion ($\rho = .59$, $p < .001$), and personal health being affected ($\rho = .56$, $p < .001$). Personal health being affected was also strongly correlated with physical strain ($\rho = .56$, $p < .001$). Together, these findings indicate that greater physical exhaustion is associated with greater mental exhaustion, worse personal health, and the increased physical strength needed for caregiving.

Physical exhaustion was moderately correlated with financial concerns ($\rho = .35$, $p < .001$), worries about the future ($\rho = .41$, $p < .001$), and ability to care for others ($\rho = .35$, $p < .001$). Mental exhaustion showed moderate correlations with the physical strength needed for care ($\rho = .44$, $p < .001$), worries about the future ($\rho = .45$, $p < .001$), and personal health being affected ($\rho = .46$, $p < .001$). Financial concerns were moderately correlated with personal health being affected ($\rho = .42$, $p < .001$), the physical strength needed for care ($\rho = .32$, $p < .001$), and ability to care for others ($\rho = .37$, $p < .001$). Personal health impacts were moderately associated with worries about the future ($\rho = .40$, $p < .001$) and ability to care for others ($\rho = .31$, $p < .001$). The physical strength needed for care was also moderately correlated with worries about the future ($\rho = .40$, $p < .001$) and ability to care for others ($\rho = .42$, $p < .001$), while worries about the future was moderately correlated with ability to care for others ($\rho = .41$, $p < .001$).

Weaker but significant correlations were observed between mental exhaustion and ability to care for others ($\rho = .29$, $p < .001$) and financial concerns ($\rho = .17$, $p = .011$), as well as between financial concerns and worries about the future ($\rho = .24$, $p < .001$).

Overall, caregiver burden domains were consistently interdependent, with physical exhaustion, mental exhaustion, and personal health impacts forming the strongest cluster, while financial concerns and future worry were related but comparatively less central. Given the correlational data and the high internal consistency of the scale (Cronbach's $\alpha = 0.82$), a caregiver burden score was calculated for each respondent as the mean of available items, with higher scores indicating greater burden. The caregiver burden index ranged from 1 to 5 ($N = 245$, $Mean = 2.56$, $SD = .85$).

Over half of respondents indicated they had no other support to care for their affected family member with DM (56%, 147/262), which included paid or unpaid support. On average, caregivers indicated they had breaks from caregiving once or twice a month ($N = 205$, $Mean = 3.15$, $SD = 1.55$). However, the responses were fairly distributed, with 26% of caregivers responding they had breaks daily (53/205) and 28% responding they never had a break (57/205).

Linear regression, shown in Table 3, was used to examine how caregiver age, level of care provided, number of people cared for, ability to take breaks, and availability of other support predicted the caregiver burden index. About 15% of the variance in caregiver burden is explained by these predictors (R-squared: 0.146), which is a moderate explanatory fit, and the model overall is statistically significant (F-test: $p = 0.0028$).

When caregiver age was examined using the 30–39 age group as the reference, the coefficients for older age groups were negative but not significant ($p > 0.1$), indicating that caregiver age does not significantly predict caregiver burden once other variables are included.

When comparing the level of care provided, there is a significant increase in burden from minor levels of care to moderate levels of care (0.347, $p = 0.024$) and to major levels of care (0.567, $p = 0.002$). This indicates that a higher level of care is associated with higher caregiver burden.

When the number of individuals with DM for whom care is provided by the caregiver was examined, it was found that caregiver burden is not significantly influenced by this factor within the context of the current model.

When comparing the role of caregiver breaks, there is an increased burden going from daily breaks to only having breaks once or twice a year (0.447, $p = 0.005$) or never having breaks (0.422, $p = 0.010$). However, there is not a significant difference between having breaks daily and having them weekly or bi-weekly. This indicates that fewer breaks are associated with higher caregiver burden for those who rarely or never get breaks.

Interestingly, the availability of additional support was not a significant predictor of burden in the regression model. This may reflect variability in the quality, consistency, or adequacy of support, suggesting that the presence of support alone may be insufficient to alleviate caregiver strain.

Table 3
Linear regression of caregiver burden index

Caregiver burden	Coef.	St.Err.	t-value	p-value	[95% Conf	Interval]	Sig
Age group (ref: 30–39)	0	.	.	.	.	.	
40–49	− .205	.372	-0.55	.582	− .938	.528	
50–59	− .491	.353	-1.39	.166	-1.186	.205	
60–69	− .503	.338	-1.49	.139	-1.171	.164	
70+	− .526	.351	-1.50	.135	-1.219	.166	
Level of assistance (ref: Minor)	0	.	.	.	.	.	
Moderate	.347	.153	2.27	.024	.045	.649	**
Major	.567	.184	3.09	.002	.205	.93	***
Number of people cared for (ref: 1 person)	0	.	.	.	.	.	
2 people	− .079	.139	-0.57	.57	− .353	.195	
3 people	.007	.19	0.04	.972	− .368	.382	
4 + people	.2	.283	0.71	.479	− .357	.758	
Breaks (ref: Daily)	0	.	.	.	.	.	
Once a week	.307	.195	1.57	.118	− .079	.692	
Once or twice a month	.247	.184	1.34	.181	− .116	.611	
1–2 times a year	.447	.156	2.86	.005	.139	.754	***
Never	.422	.162	2.61	.01	.103	.741	***
Other support available (ref.: No)	0	.	.	.	.	.	
Yes	.054	.141	0.38	.703	− .224	.331	
Constant	2.448	.367	6.67	0	1.723	3.172	***
Mean dependent var	2.673		SD dependent var	0.804			
R-squared	0.146		Number of obs	194			
F-test	2.506		Prob > F	0.003			
*** p<.01, ** p<.05, * p<.1							

Caregiver burden	Coef.	St.Err.	t-value	p-value	[95% Conf	Interval]	Sig
Akaike crit. (AIC)	464.415		Bayesian crit. (BIC)		513.433		
*** p<.01, ** p<.05, * p<.1							

When asked to select the biggest challenges they faced as caregivers in helping someone with DM, the most common responses, with nearly half responding, was dealing with emotional or behavioural changes (51%, 157/306) or supporting daily activities or mobility (42%, 128/306). Other major challenges reported included: lack of time or caregiver burnout (32%, 98/306); coordinating multiple appointments or specialists (32%, 98/306); lack of resources or support services (31%, 96/306); and communicating with healthcare providers (30%, 92/306). Respondents also indicated challenges with financial strain or insurance issues (25%, 77/306); understanding the condition and care needs (22%, 67/306); managing medications and treatments (18%, 56/306); and transportation or access to care (17%, 51/306).

Discussion

In DM, caregiver responsibilities often extend beyond physical assistance and healthcare coordination to include compensating for cognitive and behavioural symptoms such as executive dysfunction and apathy. These symptoms may increase caregivers' supervisory responsibilities and contribute to the persistent nature of caregiving demands, even in cases when physical impairment appears less severe. Caregivers of individuals with DM must manage the physical demands associated with a progressive, multisystemic disorder while simultaneously navigating the psychological impact of cognitive, behavioural, and emotional changes in their loved ones. Given the related stressors in DM care, it is perhaps unsurprising that caregivers in the study report significantly higher emotional, mental, and physical exhaustion than other burdens. For example, in an open-ended response, one spouse caring for their partner shared, "[I'm] frustrated with the apathy my husband has toward his situation and the denial that his muscular atrophy is getting worse. There are cognitive issues, but he insists he has no cognitive impact from DM." Such accounts illustrate the pervasive impact of caregiving on both mental and physical well-being.

The consistent pattern across Wilcoxon tests indicates that mental and emotional exhaustion are the most salient aspects of caregiver burden, exceeding all other domains. Caregivers particularly struggle to address the emotional changes associated with DM, which often interact with the physical challenges of care. One parent described this strain in an open-ended response, "For me, as a caregiver and parent, the emotional struggle is what takes a toll on me… I don't know how long I'll be able to assist in my sons' care. I worry about their living situation if something were to happen to me". Similarly, a parent caring for their child noted,

My daughter's affect and motivation have changed dramatically, and it is difficult to determine causative factors of her lack of follow-through. We know fatigue is a big issue but are unclear about the cognitive

changes and to what degree they impact her. I want to encourage her to do all that she can, but do not want to cause frustration and angst if she cannot do certain things.

Physical exhaustion also ranks highly and is significantly greater than most other burdens. The physical demands of DM care further heightened the emotional stress, as caregivers must adapt to progressive mobility limitations and evolving care needs. One caregiver explained,

“the number one challenge I face as a caregiver is the progression of my mother's symptoms. What may be a non-issue today will become an issue in the future... Public accessibility for those with mobility limitations are majorly lacking… Finding instruments for assistance with things like getting in and out of a car, opening a jar or drink, using an umbrella with a walker, etc. are difficult”.

This account highlights how the physical demands of DM caregiving are closely connected with emotional stress, amplifying a burden that underscores the need for comprehensive support strategies.

The Spearman correlations show that the seven aspects of caregiver burden (physical exhaustion, emotional or mental exhaustion, financial challenges, personal health issues, physical strength, future concerns, and the ability to care for other family members) are not independent but highly interrelated, particularly among physical exhaustion, mental exhaustion, and health impacts. The strong correlations ($\rho = .56–.65$, all $p < .001$) indicate a core cluster of burden, where increases in challenges in one area often exacerbate difficulties in others. This reinforces the idea that caregiver burden operates as a multidimensional but unified construct, rather than as isolated challenges. Greater attention is needed to the caregiving side of addressing the complex needs of DM caregivers. As one caregiver explained in an open-ended response,

The care burden is endless and multifaceted (medical, behavioural, cognitive)… we are also case managers and advocates… all while simultaneously providing direct care for very medically fragile and cognitively impacted family members at all hours of the day and night. We are invisible in plain sight.

The interconnection of physical, emotional, and practical demands amplifies overall burden, underscoring the critical need for research to identify effective strategies and resources that support this population, ensuring both caregiver well-being and the continuity of care for individuals living with DM. Programs designed to support caregivers must take a holistic approach that addresses the multiple, overlapping stressors they face. For example, interventions that focus only on financial assistance may miss the most impactful dimensions of burden if they do not also address emotional and physical fatigue.

Addressing caregiver burden requires both recognizing the challenges caregivers face and providing practical support that sustains their well-being. The strongest predictors of higher caregiver burden are the intensity of care required and the absence of breaks. The pronounced differences between caregivers with occasional breaks versus those with no or almost no breaks highlight the importance of policies and programs that prioritize meaningful respite, to effectively reduce stress and promote well-

being. Meaningful respite goes beyond brief or infrequent pauses; it involves sufficient, reliable periods of support that enable caregivers to rest, recharge, and attend to personal needs without worry. As one caregiver explained: “Caring for my spouse as an unaffected caregiver can be emotionally and physically taxing. I am at my best when I remember to schedule a bit of personal time for myself, as I am able, in order to recharge and rest.” Another caregiver shared their desire for more significant breaks “It would be great to get more help and support… to be able to take three or four days off would do wonders for me.” To be truly restorative, respite must also relieve caregivers of the ongoing cognitive and organizational responsibilities they frequently manage for their loved one, including healthcare coordination, appointment scheduling, medication management, prompting daily activities, and monitoring safety or follow-through. Although our model accounts for about 15% of the variance in caregiver burden, other unmeasured factors likely contribute, reflecting the complex and multifactorial nature of caregiver stress.

Overall, these findings underscore that meaningful, holistic support is essential to reducing caregiver stress and promoting well-being. Programs that provide structured, predictable, and substantial relief may significantly mitigate caregiver burden, improve mental and physical well-being, and help sustain long-term caregiving capacity. Ultimately, ensuring caregivers have access to meaningful respite and supports is a critical step in supporting their health and sustaining the care of individuals living with DM.

Study limitations

While this study is one of the largest DM-specific caregiver survey studies to date and provides new insights into their needs, several limitations should be noted. First, income and personal finances were not directly assessed. It is possible that there may have been a sampling bias of respondents, if those who were able to access and complete an online survey were more affluent than the general DM population. As this is primarily an online survey, this may introduce a bias due to technology access or digital literacy. This may mean that caregivers in the broader DM population experience even greater financial stress or other burdens than are reported here.

Second, a low percentage of respondents reported caring for more than one person. Given the genetic nature of DM, multiple family members often require care, so the survey sample may not fully reflect the broader caregiver population and may underrepresent more burdened caregivers. It is possible that caregivers responsible for multiple individuals were less able to complete the survey, which could obscure potential effects of caring for more than one person. Caregivers who are the most overwhelmed and have the least ability to respond may be the least likely to respond.

Third, few respondents reported being affected by DM themselves. A concurrent survey was administered to individuals with DM. People affected by the disease may have selected to only fill out that survey, as a result we may be missing out on those who are affected by the disease and also caregiving, perhaps for a child or grandchild. As a result, this study may underrepresent caregivers who are managing their own disease in addition to caring for others living with the disease.

The survey relied on self-reports of experiences, as such there may be self-report or recall bias. For example, some caregivers may underreport strain due to a desire to appear to be coping well. Or if caregivers were distressed at the time of completing the survey, they may overreport their burden. Additionally, because most caregivers supported individuals with DM1, the study may have limited power to characterize DM2-specific caregiving experiences.

Denominators varied across survey items because of missing responses and skip patterns, and regression analyses used complete cases only. The caregiver burden index demonstrated good internal consistency in this sample, but it was constructed from adapted items and should be interpreted as a study-specific measure rather than a fully validated DM caregiver burden instrument. Because the data are cross-sectional, associations between care intensity, respite frequency, and burden should not be interpreted causally.

There may be additional caregiving burdens not captured in this survey. The study used a cross-sectional, self-reported survey distributed primarily through MDF-related channels, including conference attendees, email lists, social media, website links, and the Myotonic Dystrophy Family Registry. As a result, respondents may represent a more engaged, resource-connected, English-speaking, and predominantly US-based caregiver population than the broader DM caregiver community. The sample was also predominantly white, female, and over age 50, which may limit generalizability to more diverse caregiver groups. The findings may not generalize to caregivers from underrepresented racial/ethnic groups, non-English-speaking caregivers, caregivers outside the US/Canada, or those not connected to MDF or registry networks. Taken together, these limitations suggest that the overall burden of caregiving in the general DM population may be even greater than observed, highlighting the significance of the challenges documented in this study.

Given many caregivers are parents and or guardians and some are over 70, the sample likely includes caregivers of adults with congenital or childhood-onset DM1. That group may have very different burden than caregivers of adult-onset DM1 or DM2. Future work should examine caregiver burden by disease subtype, age of onset, relationship to the affected individual, and functional severity.

Conclusions

This study provides a comprehensive examination of the unique challenges faced by caregivers of individuals with DM1 and DM2. These findings demonstrate that caregiving in DM may differ from that in many other chronic and neuromuscular conditions due to the overlapping effects of progressive physical decline and cognitive-behavioural involvement, which together create a uniquely demanding and ongoing caregiving burden. The findings highlight that caregiving in DM is multidimensional, encompassing emotional and psychological strain, physical exhaustion, financial pressures, and substantial healthcare coordination responsibilities. These burden domains are interconnected, suggesting that caregiver stress in DM operates as a cumulative and reinforcing experience rather than as a series of isolated challenges. Caregivers take on complex roles as advocates, coordinators, supervisors, and long-term

care providers while also managing their own health, aging, finances, and future caregiving capacity. Caregiver burden is closely tied to the intensity of care and limited access to meaningful respite, highlighting the need for sustained, high-quality, and integrated support models rather than fragmented or short-term solutions. These findings underscore a clear need for targeted support interventions, including opportunities for respite, particularly for those providing high levels of care, as well as broader, comprehensive resources to address the diverse challenges caregivers encounter. Overall, these findings underscore the need to recognize caregiver support as a central component of comprehensive DM care, including the development of DM-specific supports that address the sustained physical, cognitive, emotional, and organizational demands placed on caregivers over time. Future research should include longitudinal studies to better understand how caregiver burden evolves across the disease course, as well as interventional studies evaluating the effectiveness of respite and caregiver support programs. Future work should examine caregiver burden by disease subtype, age of onset, relationship to the affected individual, and functional severity. Additional research is also needed to better understand the experiences and support needs of affected caregivers, whose dual roles as both caregiver and individual living with DM may present distinct challenges that remain underexplored.

Abbreviations

DM: Myotonic Dystrophy

DM INSIGHT: Investigating Needs, Support, Gaps, and Healthcare Trends in Myotonic Dystrophy

MDF: Myotonic Dystrophy Foundation

NMD: Neuromuscular disease

NORD: National Organization for Rare Disorders

RDDC: Rare Disease Diversity Coalition

Declarations

Ethics approval and consent to participate

The DM INSIGHT study was approved by the Advarra Institutional Review Board under the following protocols: Myotonic Dystrophy Foundation – DM INSIGHT: Investigating Needs, Support, Gaps, and Healthcare Trends (Canada) (Pro00086366) and Myotonic Dystrophy Foundation – DM INSIGHT: Investigating Needs, Support, Gaps, and Healthcare Trends (Pro00086144). All participants provided informed consent prior to participation in the study. Consent was obtained electronically at the start of the survey, and participants were informed of the study purpose, procedures, risks, and their right to withdraw at any time without consequence.

Availability of data and materials

The datasets generated and/or analysed during the current study are not publicly available due to privacy and confidentiality restrictions but are available from the corresponding author on reasonable request. Survey instruments are available at myotonic.org.

Competing interests

The authors are employees or representatives of the Myotonic Dystrophy Foundation, which funded and conducted the study.

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Authors' contributions

All authors contributed to the conceptualization and design of the study, development of the study protocol and survey instruments, and drafting and critical revision of the manuscript. NS conducted all statistical analyses and interpreted the data. MB led the pilot testing and facilitated the focus groups. TS provided overall study oversight and senior leadership. All authors reviewed and approved the final manuscript.

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