the christopher project

Report to the Myotonic Dystrophy Community

Prepared by: Sarah Howe, Marigold Foundation and the Christopher Project Reference Group | Spring 2019

Acknowledgements

As a collaborative effort, the success of the Christopher Project has drawn upon the contributions of many different people and groups. We are grateful for their willing participation and support:

- The Association Française contre les Myopathies (AFM) and their 2010 study of patients and families living with myotonic dystrophy in France
- Project Partners (listed alphabetically): Groupe de reserche interdisciplinaire sur les maladies neuromusculaires (GRIMN), Marigold Foundation, Muscular Dystrophy Association (MDA), Muscular Dystrophy Canada (MDC), Myotonic Dystrophy Foundation (MDF), Stanford School of Medicine, and University of Rochester Medical Center (URMC)
- Project Reference Group Members (past and present, listed alphabetically): Diane Bade, Marie-Hélène Bolduc, Elizabeth Florence, Paul Formaker, Cynthia Gagnon, Katharine Hagerman, Chad R. Heatwole, Leah Hellerstein, Sharon Hesterlee, Sarah Howe, Don MacKenzie, Sue McIntyre, Lianna Orlando, John Porter, Marla Spiegel, Kristin Stephenson, Eric T. Wang, and Jodi Wolff
- Dwayne Dobson at Combine Design & Communications and Dave Hibbeln at H-one Design
- Numerous industry, regulatory, and government representatives

And a special thank you to the patients and families across the United States and Canada who participated in the Project, and all of the "Christophers" among them. This report is for you!

The Christopher Project was named after a real person living with myotonic dystrophy in the American Midwest.

He has a loving family and a good doctor who do their best but sometimes Christopher faces challenges in managing his myotonic dystrophy that are difficult to overcome.

We can all relate to Christopher –
he could be any one of us.

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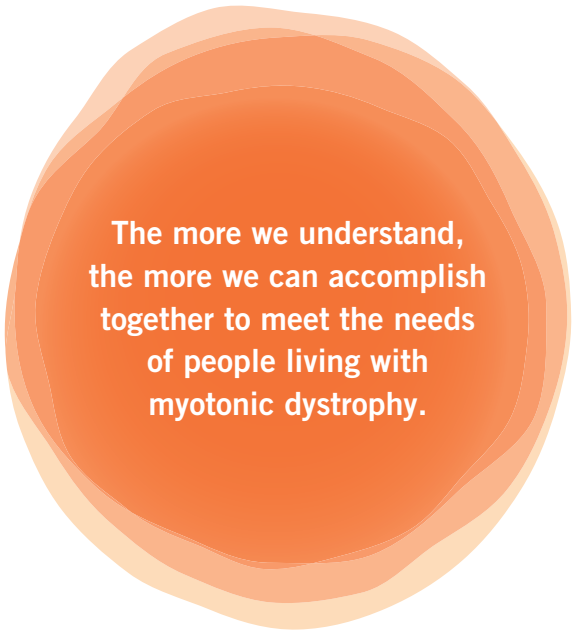
Introduction

Myotonic dystrophy is a complex neuromuscular disorder. It affects many different parts of the body in many different ways. It is also rare: most people have never heard of it and many physicians and healthcare providers may not encounter a patient with the condition or recognize it when they do (Harper, 2002). Each person living with myotonic dystrophy has their own unique experience. For many, the need for timely and accurate information, competent medical care, and appropriate treatments and therapies often goes unmet.

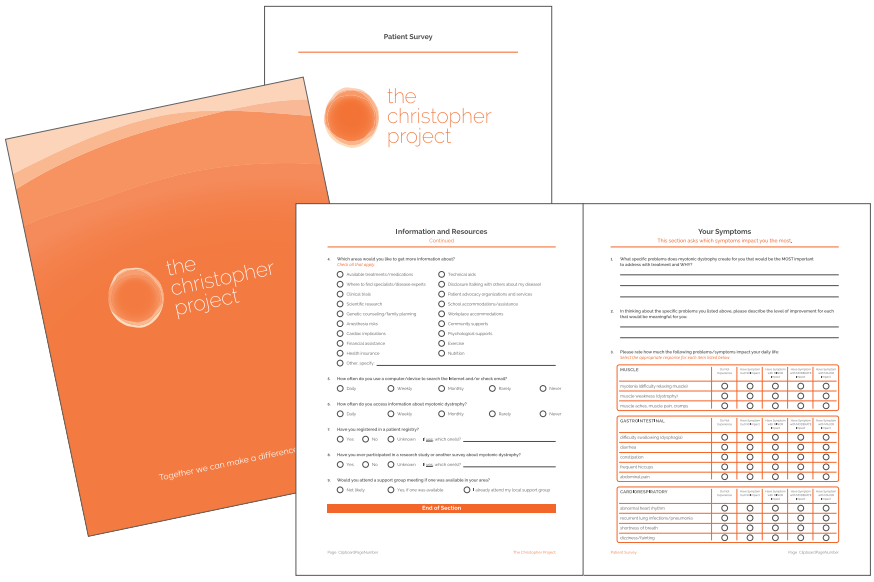
In order to better understand their needs, the Christopher Project asked patients with myotonic dystrophy and their family members/caregivers to report directly on their own experiences, from their own perspective, as experts in their own right.

Patients and family members/caregivers who participated in the project provided a wealth of information about their diagnostic experience, symptoms, daily life challenges, and healthcare-related experience. The significant input they provided, which is summarized in this report, complements existing research and offers important, new insight into the myotonic dystrophy experience. It will serve as an invaluable and lasting resource for the benefit of the entire community.



Methodology

The Christopher Project is an important research collaboration between national patient advocacy organizations, healthcare providers, patients, and family members/caregivers in the United States and Canada. It set out to capture the diverse experiences of people living with myotonic dystrophy directly from patients and family members/caregivers.



In order to accomplish this, a large patient survey with over 150 questions was distributed in the summer of 2014 to almost 4,000 individuals that had been randomly selected from current advocacy group mailing lists. Survey questions included a broad range of topics covering daily life, current health, and healthcare-related experiences. Individuals were asked to fill out the survey on their own (or with help if needed) and mail it back for data entry.

In early 2015, a follow-up survey with about 100 questions was distributed to 1,000 family members/caregivers to learn about their experiences and to gather additional insight into the reported patient experience.

1,180 complete patient surveys (30% response rate) and 402 complete family member/caregiver surveys (40% response rate) were received, coded, and anonymized to safeguard privacy. Over 200,000 data points from these surveys were then entered into a secure database and a rigorous process of descriptive analysis was performed. This report presents the outcomes of this analysis.

About this Report

This Report to the Community is for anyone who wants to know more about what it is like to live with myotonic dystrophy. For ease of access, the considerable amount of information contained in this report is organized into the following major sections:

Section 1: *Key Findings* — *A summary of the major findings of the Christopher Project and a discussion of their implications.*

Section 2: *Opportunities for Moving Forward* — *Identification of areas for action for various stakeholders that follow directly from the key findings.*

Section 3: *Patient Survey Results* — *Detailed results of the entire patient survey. Includes results for all respondents and for each specific type of myotonic dystrophy.*

Section 4: *Family Member/ Caregiver Survey Results* — *Detailed results of the entire family member/caregiver survey. Includes results on their own experience and their view on the patient experience.*

This report gives voice to the many patients and family members/caregivers who completed the original Christopher Project surveys. It is not intended to replace expert medical advice nor prescribe changes in clinical care. Instead, it is a useful reference that anyone living with myotonic dystrophy can use to:

- Learn more about the myotonic dystrophy experience.
- Gain additional perspective on their own experience.
- Help advocate for the information, care, and support they need.

Additional copies of this report can be found online at: www.christopherproject.org

Some of the results of the Christopher Project have also been published in the scientific journal Muscle & Nerve:

Hagerman KA, Howe SJ, Heatwole CR; Christopher Project Reference Group. The myotonic dystrophy experience: a North American cross-sectional study. *Muscle Nerve*. 2019 Jan 24. doi: 10.1002/mus.26420.

About the Different Types of Myotonic Dystrophy

Everyone living with myotonic dystrophy has a specific type of the disease. Throughout this report, certain findings are presented by these different type groups as described below:

Myotonic Dystrophy (Dystrophia Myotonica, DM)

Myotonic dystrophy is a highly variable, multi-systemic, progressive neuromuscular disease. The age of symptom onset can occur from birth throughout any stage of life, and muscle and other symptoms can be minimal to severe. There are two main types of the disease: myotonic dystrophy type 1 (DM1; also called Steinert's Disease) and myotonic dystrophy type 2 (DM2; historically referred to as Proximal Myotonic Myopathy (PROMM)). Though similar in presentation, each type has a different genetic cause.

Myotonic dystrophy type 1 can also be broken down into subtypes: congenital (cDM1), infantile, juvenile, adult, and late-onset (De Antonio, et al, 2016). For the purposes of this report, cDM1 is reported on separately and the other subtypes are included in the DM1 report.

Myotonic Dystrophy Type 1 (DM1)

Age of onset: Early childhood through adulthood

Symptoms: Mild DM1 is characterized by cataracts before age 50 and myotonia (difficulty relaxing muscles). Classic DM1 is characterized by muscle weakness, muscle wasting, myotonia, sleep and other cognitive issues, gastrointestinal complications, early cataracts, and often cardiac conduction abnormalities.

Congenital Myotonic Dystrophy Type 1 (cDM1)

Age of onset: Birth

Symptoms: Low muscle tone and severe overall weakness at birth, resulting in feeding and/or breathing problems that can require intervention and intensive care; intellectual disability is common.

Myotonic Dystrophy Type 2 (DM2)

Age of onset: Typically mid-adulthood

Symptoms: Myotonia (difficulty relaxing muscles), muscle pain, muscle weakness, sleep disturbances, gastrointestinal issues, cardiac conduction defects, and cataracts before age 50. There is no known severe childhood form of DM2.

A detailed overview of the different types of myotonic dystrophy is available online at GeneReviews: www.ncbi.nlm.nih.gov/books/NBK1165 and www.ncbi.nlm.nih.gov/books/NBK1466

Section 1: Key Findings

The findings of the Christopher Project represent a broad sample of male and female patient and family member/caregiver respondents from all regions of the United States and Canada, from all age groups, across all types of the disease.

Respondents demonstrated a deep understanding of their disease, its breadth of symptoms, and its impact on their daily lives. They provided vital perspective on their medical needs and healthcare-related experiences. They identified areas where they are successfully managing and areas where they are having challenges. Key findings on their reported experiences are summarized as follows:

Diagnosis

Patients reported a wide range of experiences around their diagnosis:

- Myotonic dystrophy symptoms can begin at any age and stage of life.
- The average age of onset differed dramatically between different disease 'types'.
- Many respondents experienced a significant time delay between onset of symptoms and their diagnosis. The length of this delay varied across different disease 'types'.

The support provided at the time of diagnosis is not sufficient:

- A significant number (21%) of patient survey respondents received 'no assistance' when they were diagnosed.
- Only 21% received genetic counseling when they received their diagnosis.
- More than half of patient respondents said the emotional impact of receiving their diagnosis was a specific challenge yet only 6% received psychological support.

Each person diagnosed with myotonic dystrophy has their own unique experience and diverse set of needs. Proper support, information, and guidance is essential at this critical time.

More Key Findings >>>

Type of Myotonic Dystrophy (cDM1, DM1, DM2)

The experiences of patient respondents varies across the different 'types' of the disease:

- **cDM1** respondents were significantly younger and more dependent on their family members/caregivers and healthcare providers compared to adult onset **DM1** and **DM2**. They were more impacted from psychological symptoms like learning difficulties and had greater difficulty concentrating. They reported greater challenges performing daily activities than the **DM1** and **DM2** groups.
- **DM1** respondents reported more daytime sleepiness and more impact from fatigue, and a harder time staying alert than **cDM1** and **DM2** respondents. **DM1** respondents were also more affected by gastrointestinal symptoms, had more myotonia, and had more difficulty handling objects than the other type groups.
- When compared to **cDM1** and **DM1** groups, **DM2** respondents were typically older. They reported a later age of onset and a longer delay between age of onset and age at diagnosis. They were more impacted by muscle pain, muscle aches/cramps, and balance issues. They reported greater mobility challenges, yet more success in school and work.

Differences observed between the type groups are important and relevant but type is not the only predictor of disease experience. Many other factors such as age, disease progression, gender, and healthcare access can contribute to these reported differences.

Many respondents are uncertain about the 'type' of myotonic dystrophy they have:

- 28% of patient respondents stated they do not know or are unsure of the 'type' of myotonic dystrophy they have (identified as unknown/unsure (**U/U**) in this report).
- This uncertainty was even present among the 70% of respondents who indicated they had received genetic confirmation of their disease.
- Family member/caregiver respondents also reported uncertainty around type.

Uncertainty around 'type' may be because genetic testing was not yet available or was not offered at diagnosis. It could also be that the patient was not told their specific type, or they were told and simply do not remember.

There is ambiguity amongst respondents who reported they have cDM1:

- Some patient respondents reported they have **cDM1** but reported an age of onset other than birth, which is not consistent with the clinical definition of **cDM1**.

This ambiguity may come from the use of the term 'congenital', which means 'to be born with' and is commonly used to describe hereditary diseases in general.

Knowing the specific 'type' of myotonic dystrophy a patient has is necessary for understanding the nature and progression of the disease, appropriate clinical care options, and relevance of potential therapies. Working directly with a qualified professional for a confirmatory diagnosis is the best way for patients and their healthcare providers to obtain this information.

Gender

There are some notable differences between male and female respondents:

- More female respondents reported greater impact on their daily life from abdominal pain, diarrhea, constipation, headaches/migraines, and dizziness/fainting compared to male respondents.
- More male respondents reported greater impact on their daily life from hair loss, learning difficulties, sleep apnea, and intimacy problems when compared to female respondents. They also reported greater challenges with certain household/personal needs, communication, social, school, and work activities compared to females.
- More female respondents lived with their spouse/partner and/or children while more male respondents lived with their parents.

While the findings for male and female respondents are closely aligned, the differences have implications for daily life, appropriate disease management, and clinical care. They are also relevant to current and future research, given the identification of gender as an influencing factor in the clinical profile and severity of myotonic dystrophy (Dogan et al, 2016).

Symptoms

Myotonic dystrophy is highly variable and affects many different parts of the body:

- Patients reported they experience a wide range of symptoms with varying degrees of impact on their daily lives.
- Muscle-related symptoms, sleep and fatigue symptoms, and balance were the most prevalent and impactful symptoms.
- Gastrointestinal issues and psychological symptoms also ranked highly in prevalence and impact.
- Symptomatology and disease severity varied greatly between patients, across different stages of disease progression, and among the different types of myotonic dystrophy.

The prevalence and impact of muscle symptoms and sleep issues reinforce existing knowledge in myotonic dystrophy. The prevalence and impact of gastrointestinal problems, balance issues, and psychological symptoms provide new insights into the patient experience because they are generally not as well understood.

This diversity and breadth of symptoms places substantial physical, psychological, and emotional burdens on patients and family members/caregivers with major implications for all aspects of daily life and clinical care.

Effective symptom management requires a comprehensive and coordinated approach across many different disciplines.

More Key Findings >>>

Managing Daily Life

Performing daily activities can be challenging:

- The ability to perform daily life activities varied greatly between individual patients and between different types of activities.
- Mobility and household/personal needs activities were the most challenging: the majority of respondents reported some level of challenge in performing these activities.
- Doctors office, school, and social activities were less challenging: one-third of respondents reported some level of challenge performing these activities, yet about half of respondents reported them as 'not a challenge'.

Getting around effectively is particularly difficult:

- Muscle-related symptoms and balance issues affected the majority of patient respondents.
- Mobility-related daily life activities were the most challenging to perform, especially going up and down stairs, standing, and maintaining balance.
- Mobility issues were identified as the 'biggest obstacle' by one-third of both patient and family member/caregiver respondents.

It is not surprising that patients face challenges in performing activities throughout their daily life given the diversity and breadth of symptoms they experience.

It is also not surprising, given the variability and progressive nature of myotonic dystrophy, that some respondents report little or no challenge with certain activities, while others report major challenges.

Patients need to be aware of their unique challenges. They need to talk about them with their family members/caregivers and healthcare providers in order to develop strategies to get the support they need and mitigate the impact these challenges have on their daily lives.

Patient respondents face major challenges in their working lives:

- While patient respondents reported education levels at or above national averages, they are employed at substantially lower rates than the rest of the general population.
- Half of patient respondents indicated at least some level of challenge with employment (obtaining/retaining work); one-quarter reported they are 'unable to perform' at work.
- When asked directly about their employment status, more than half of all respondents reported they are 'unable to work due to myotonic dystrophy'.
- Income levels reported by patient survey respondents are lower than national norms.

Employment is a key component in quality of life and critical to social, emotional, and financial wellbeing. Strategies are needed to improve employment prospects for people living with myotonic dystrophy that recognize their strengths and limitations, increase workplace awareness, and advocate for workplace accommodations.

Survey results provide only basic insight. More research is needed to adequately understand the considerable challenges people with myotonic dystrophy face in obtaining and maintaining employment.

Healthcare Experience

Respondents report positive healthcare experiences:

- Patient respondents were generally engaged with the medical system. Most reported seeing their family doctor, neurologist, and/or cardiologist regularly.
- Patient and family member/caregiver respondents are satisfied with the healthcare the patient receives. Only 10% reported any level of dissatisfaction at all.
- Medical specialists were reported as the most helpful source of information.
- 'Talking with healthcare providers' was one of the most preferred ways to receive information about myotonic dystrophy.
- 85% of patient respondents reported they 'always' or 'often' follow their doctors' advice.

Patients and family members/caregivers clearly articulated that they rely on their healthcare providers for information, care, and support. This is a positive finding and implies that many patients have established effective and trusting partnerships with their current healthcare providers. Building upon this success is key to meeting the needs of patients.

Access to different healthcare providers varies among patients:

- Primary and secondary care providers, like family doctors/GPs, neurologists, and cardiologists, were seen regularly by most patients.
- Other medical specialists, like pulmonologists and gastroenterologists, were seen less frequently.
- Allied health professionals, like speech/swallowing specialists, nutritionists, and occupational therapists, were also seen less frequently.

Every person living with myotonic dystrophy has their own unique experience: not everyone has the same healthcare needs and a person's needs change over time. As such, variable access to specific providers is expected.

There are some critical gaps in healthcare access:

- Myotonic dystrophy is a serious neuromuscular disease and 86% of respondents had seen a neuromuscular specialist/neurologist. However, 12% had never seen a neuromuscular specialist/neurologist.
- Myotonic dystrophy has serious cardiac implications and 79% of survey respondents had seen a cardiologist. However, 18% had never seen a cardiologist.

The high percentage of respondents who have seen these specialists is a positive finding. However, there remains significant percentages of people who have not. This is a serious area of unmet need given the critical support that neuromuscular specialists and cardiologists are able to provide.

More Key Findings >>>

There are other identifiable gaps in healthcare access:

- Myotonic dystrophy is a hereditary/familial disease and has serious implications for family planning, yet 72% had never seen a genetic counsellor.
- 44% of respondents who reported major impact from gastrointestinal issues had never seen a gastroenterologist.
- 49% of respondents who reported major impact from sleep symptoms had never seen a sleep specialist.
- 53% of respondents who reported major impact from cognitive symptoms had never seen a psychologist or a psychiatrist.

These gaps speak directly to the challenges of effectively managing a complex, multi-systemic disease and represent areas where improved access could result in better care and patient outcomes.

A comprehensive and informed system of clinical care is essential to meeting the varied medical needs of patients.

Some patient respondents did not know about certain risks associated with myotonic dystrophy:

- Nearly half of patients reported they were not aware of general anesthesia risks associated with myotonic dystrophy and/or had not discussed them with a medical professional.
- Nearly half of patients reported they were not aware of cardiac risks associated with myotonic dystrophy and/or had not discussed them with a medical professional.
- There is ambiguity around cardiac care in myotonic dystrophy and nearly one in five respondents reported they had never seen a cardiologist.

Patient and healthcare providers must be made aware of these risks so they can access relevant information and develop appropriate clinical care strategies to mitigate them.

The development of resources by myotonic dystrophy experts, such as consensus-based care recommendations (Ashizawa et al, 2018), anesthesia guidelines, and the Myotonic Dystrophy Foundation Toolkit, have helped to establish an important framework for clinical care and disease management. The ongoing development and distribution of resources like these will contribute to improved health outcomes.

More Key Findings >>>

Treatments and Interventions

There is no clear picture of what medications may be appropriate or effective:

- Only a quarter of respondents reported they took any medication to manage their myotonia (difficulty relaxing muscles), daytime sleepiness, fatigue, or gastrointestinal issues.
- Respondents reported the use of 45 different medications for myotonia, 20 for daytime sleepiness and/or fatigue, and 51 for gastrointestinal issues. The level of satisfaction with the use of these medications was mixed.

There are currently no medications specifically approved for myotonic dystrophy and there are very few clinical studies that have systematically evaluated the use of therapeutic agents in myotonic dystrophy. The lack of scientific evidence paired with the multi-systematic and highly variable presentation of the disease make the identification and selection of appropriate medications particularly challenging for prescribing physicians.

In the absence of clear scientific data, it is likely that many patients are being prescribed medications that are of limited benefit, pose unnecessary risk, or are potentially harmful. It is also possible that understudied therapies exist that could alleviate disease burden in myotonic dystrophy. There is an urgent need to identify existing medications that are potentially safe and effective for myotonic dystrophy patients and evaluate these therapies using appropriate clinical trials.

Importantly, there are currently no medications that are approved as a treatment for the root cause of myotonic dystrophy. This is perhaps the greatest area of unmet medical need for patients and families. The development of new drugs that might slow, arrest, or reverse the underlying disease processes has the potential to be life-changing.

Other available treatments and interventions are underutilized:

- Patients identified mobility as one of their greatest challenges yet the use of mobility devices, like leg/ankle braces, canes and crutches, and wheelchairs, was low.
- Although 47% of respondents reported that they have sleep apnea (trouble breathing during sleep), the use of assistive nighttime breathing devices, like CPAP and BiPAP, was low.
- Frequency of access to medical specialists and allied health professionals who can provide symptomatic care and management was low.

Greater access to specialist care, select use of available non-pharmacological treatments, and focused symptomatic management is warranted and can contribute to improved health outcomes.

More Key Findings >>>

Patients and family members/caregivers want to know more about treatments and interventions:

- Almost three quarters of patients and family members/caregivers said they want to know more about available treatments/medications.
- Half of patients and family members/caregivers said they want to know more about clinical trials.
- Half of patients and family members/caregivers said they want to know more about exercise.
- Almost half of patients and family members/caregivers said they want to know more about nutrition.

There is a need for healthcare providers and patient advocacy organizations to make the most current and relevant information on potential treatments and interventions available to patients and family members/caregivers. This is increasingly important as the field of drug development for myotonic dystrophy is more active than ever before.

Respondents do not have a clear picture of opportunities for patient participation:

- Less than half of patients reported they participate in a registry and about one quarter did not know if they were currently registered in one.
- Only 20% of patients indicated they had previously participated in a survey or research study related to myotonic dystrophy.
- Many respondents did not provide any details when asked about the specifics of their participation (i.e., registry name or research study name).

New and current opportunities for patient participation need to be promoted. Also, more can be done to inform people who are already engaged to help them better understand their participation and the value of their contributions.

Patients and family members/caregivers want more information to help them manage their myotonic dystrophy:

- 43% indicated that a lack of resources available about their disease was a major challenge at their time of diagnosis.
- 20% of patients specifically identified ‘information’ as an important area of unmet need.
- Respondents identified a long list of areas they want to know more about —the top three were available treatments/medications, clinical trials, and exercise.
- Respondents were interested and open to receiving information in many different ways—the top three most preferred were lettermail, speaking directly with healthcare providers, and reading printed materials/books.

The availability of accurate and timely information on the nature, progression, and management of myotonic dystrophy is critical. Patients and family member/caregivers also need ongoing access to relevant information on available resources, systems of support, and managing daily life.

Information needs shift and change over time and many patients and family members/caregivers may not be aware of the resources that are currently available. Satisfying these needs requires a concerted and sophisticated approach.

Many patient respondents are receiving support from family members/ caregivers at home:

- Of the 402 family member/caregiver respondents, most were spouses or parents who live with the original patient survey respondent.
- Most of them provided support across all aspects of the patient's daily life, including substantial emotional support.

Patients and family members/caregivers have to manage myotonic dystrophy day in and day out. For many patients, family members/caregivers are the main system of support outside the clinic. As such, the role of family members/ caregivers is extremely important.

Family member/ caregiver respondents face significant challenges:

- Nearly 1 in 5 family member/caregivers reported they also have myotonic dystrophy themselves.
- Over a quarter provided support to more than one person affected by myotonic dystrophy.
- Family members/caregivers reported that managing the complexity of the disease, the burden on themselves as a caregiver, and efforts to play a coaching/supporting role are their greatest challenges.
- Over one-third specified they would like to know more about caregiver support and training.

The impact of myotonic dystrophy on family members/caregivers is substantial and should not be overlooked. Healthcare professionals, clinical centers, and advocacy organizations have an opportunity to develop targeted initiatives to address the specific needs of those caring for people with myotonic dystrophy.

Patients and their family members/ caregivers share a common understanding of what it's like to live with myotonic dystrophy:

- Patients and family members/caregivers provided very similar responses across all comparative categories and questions (i.e., symptom prevalence and impact, daily life challenges, patient general health, disease type, first symptoms, age of onset, compliance, satisfaction with healthcare, obstacles, and unmet needs).
- There were only a few differences. In those instances, family members/ caregivers tended to report that patients were slightly more impacted by myotonic dystrophy than the patients themselves reported.

This major alignment is a positive finding. It demonstrates that family members/caregivers understand what patients are going through and provides a strong basis for them to work together towards positive health outcomes.

Importantly, this close alignment from two different perspectives provides strong evidence of the reliability of the patient findings presented throughout this report.

– End of Section 1 –

Detailed results for all survey questions can be found in this report in *Section 3: Patient Survey Results* and *Section 4: Family Member/Caregiver Survey Results*.

Section 2: Opportunities for Moving Forward

The findings of the Christopher Project improve our understanding of the needs of people living with myotonic dystrophy, where they are being met, and where they are not being met. As such, they point to opportunities where various stakeholders can play an active role to better meet these needs.

Opportunities for Patients

Patients should be at the center of any effective disease management strategy and should take an active role to ensure they get the information, care, and support they need.

Patients need to understand what type of myotonic dystrophy they have. They can speak directly with a qualified healthcare provider to get this information and understand what it means for disease management and progression. They can also request genetic testing for confirmation.

Patients need access to timely and relevant information about myotonic dystrophy so that they understand their disease and their medical needs. They can speak directly with qualified healthcare providers, engage with patient advocacy groups, talk with family members, and do their own research to educate themselves.

Patients need to engage with healthcare providers and should advocate for their own care. They should develop a partnership with a primary healthcare provider (normally a family physician or a neuromuscular specialist/neurologist) who can help them coordinate referrals to specialists and provide timely follow-up. They should prepare for clinical visits, take the time to discuss their needs with their healthcare providers, and follow through on their providers' recommendations.

Patients need to advocate outside the clinic too. They should work together with family members/caregivers, support groups, teachers, and employers to develop appropriate strategies, tools, and accommodations that address their needs between clinical visits.

Patients can contribute more broadly by joining patient registries and participating in research including clinical drug trials.

Opportunities for Healthcare Providers

Healthcare providers play a crucial role in ensuring patients get the information, care, and support they need.

Healthcare providers need to provide patients with an accurate and timely diagnosis (including the specific type of myotonic dystrophy). They should deliver it in a way that provides appropriate guidance, relevant information, and fundamental support at this very challenging time.

Healthcare providers need to rely on the best of their knowledge, skills, and abilities to understand and manage the multi-systemic, highly variable, and progressive nature of myotonic dystrophy. They can also draw upon the wealth of experience patients and family members/caregivers bring to the clinic.

Healthcare providers should partner with patients to develop a comprehensive system of care that addresses all aspects of the disease. This system of care should meet patients' current needs, be responsive to the progressive nature of the disease, and include appropriate referrals to specialists, necessary screening, and coordinated follow-up.

continues >>>

Healthcare providers are relied upon to provide accurate, timely, and relevant information to patients and family members/caregivers to help them manage their disease. They should take the time to talk directly with patients and family members/caregivers and answer questions, provide printed materials/handouts, and recommend additional information and resources.

Healthcare providers can contribute to the vitality of the broader community by participating in patient-centered activities, promoting patient advocacy, and facilitating patient-centric research.

Leaders in the field can work together with other experts and advocacy groups to develop and disseminate standards of care and information and resources for the benefit of the broader community.

Opportunities for Scientists and Clinical Researchers

Basic scientists can direct their research to address underlying disease mechanisms that are directly connected to what matters most to patients and family members/caregivers.

Basic scientists and clinical researchers can further expand their research initiatives to address additional clinically important aspects of myotonic dystrophy such as central nervous system (CNS) deficits, gastrointestinal issues, and cardiac complications.

Clinical researchers can actively work together to gather evidence regarding medications, devices, and therapies that are suitable for use in myotonic dystrophy and disseminate this information broadly for the benefit of the entire community.

Clinical researchers can work together with industry to facilitate clinical drug trials that address the areas that are relevant and meaningful to patients and family members/caregivers.

Opportunities for Biotechnology and Pharmaceutical Companies

There are a considerable number of opportunities for therapeutic intervention due to the multi-systemic nature of myotonic dystrophy. Biotechnology and pharmaceutical companies can pursue approaches aimed at treating individual symptoms and/or targeting underlying disease mechanisms.

Biotechnology and pharmaceutical companies should develop treatments that are relevant and meaningful to patients. They can inform their drug development programs by partnering with expert scientists and clinical investigators, engaging with patient advocacy organizations, and listening closely to patients and family members/caregivers about what matters most.

Opportunities for Government and Regulatory Bodies

Government bodies need to understand the considerable burden myotonic dystrophy places on patients, families, and the healthcare system across multiple domains and provide funding accordingly.

Regulatory bodies need to recognize the multi-systemic nature of the disease and the substantial unmet medical needs of myotonic dystrophy patients when considering potential treatments and therapies.

Opportunities for Patient Advocacy

Patient advocates and advocacy organizations play a critical role in helping patients get the information, care, and support they need.

Patient advocates and advocacy organizations are an ideal source for timely and accurate information for patients, family members/caregivers, and healthcare providers on available treatments and therapies, best practices in healthcare, and strategies for managing daily life.

Patient advocates and advocacy organizations need to continue to work together with healthcare providers to enable patient access to appropriate clinical care. They can help to promote best practices and care guidelines based upon the considerable expertise that exists within the myotonic dystrophy community.

Patient advocates and advocacy organizations can enable patient access to services and support. They need to work closely with patients and families to match them with available programs, equipment, and other resources outside the clinic.

Patient advocates and advocacy organizations represent the collective voice of patients and their families at local, state, and federal government levels. They need to continue to raise awareness and press for increased funding, programs, and improved access to services and care.

Patient advocates and advocacy organizations need to advocate with industry, government, and regulatory bodies for the development and approval of meaningful treatments. They also help to facilitate patient participation in registries, clinical research studies, and drug trials.

Importantly, patient advocates and advocacy organizations are a bridge.
They strengthen the whole myotonic dystrophy community.

For more information, go to:

Muscular Dystrophy Association: www.mda.org
Myotonic Dystrophy Foundation: www.myotonic.org
Muscular Dystrophy Canada: www.muscle.ca

The opportunities identified above represent some of the areas where the myotonic dystrophy community can continue to make positive strides towards meeting the needs of people living with myotonic dystrophy. Additional and ongoing research into the patient experience will undoubtedly point to further opportunities for moving forward.

– End of Section 2 –

Section 3: Patient Survey Results

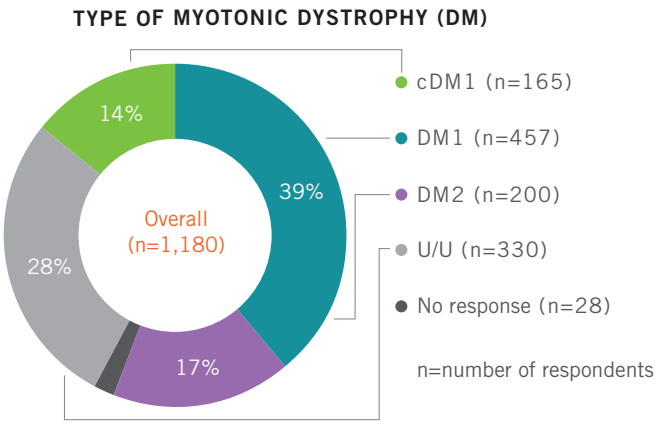
This section includes detailed results from the 1,180 overall respondents who completed the Christopher Project Patient Survey in Summer 2014.

Results by Type

Survey respondents were asked to identify which 'type' of myotonic dystrophy they have in order to understand the unique experiences of people living with each of the different types (as described in the *Introduction*).

Type of Myotonic Dystrophy Reported

As shown below, the largest group of respondents reported they have myotonic dystrophy type 1 (**DM1**), followed by unknown/unsure (**U/U**), myotonic dystrophy type 2 (**DM2**), and congenital myotonic dystrophy type 1 (**cDM1**). A small number of respondents did not provide a response to this question (shown in dark grey).



Reporting Results by Type

The combined results for all survey respondents are represented throughout this report as:

- **Overall in orange**

Additionally, results for each specific type of myotonic dystrophy are represented throughout this report by their own distinct color for easy reference:

- **Congenital myotonic dystrophy type 1 (cDM1) in green**
- **Myotonic dystrophy type 1 (DM1) in teal blue (includes infantile, juvenile, adult, and late-onset)**
- **Myotonic dystrophy type 2 (DM2) in purple**

Reporting Results by Type *continued*

The presentation of survey results by type allows readers to relate their own personal experiences to the experiences of others. It also allows those who may be uncertain about their specific type to find affinity with the experience of those in a specific type group.

Importantly, this report is not meant to be a diagnostic tool. Respondents or readers who are interested in understanding more about their specific diagnosis and/or the different types of myotonic dystrophy should speak with a healthcare professional.

A Note About the cDM1 Group

People with congenital myotonic dystrophy type 1 are born with significant symptoms, as described in the *Introduction*. Some respondents who indicated they have congenital myotonic dystrophy type 1 reported onset of symptoms later in life, suggesting they may actually have **DM1** with infantile, juvenile, adult, or late-onset.

In order to highlight the significant challenges of those living with the more severe form of the disease, only those respondents (n=71) who reported having congenital myotonic dystrophy type 1 and an age of onset at birth and received their diagnosis in the first two years of life are presented in the **cDM1** group throughout this report. The remaining respondents (n=94) are included in the **Overall** group.

Uncertainty Around Type

28% of respondents (n=330) reported they do not know or are unsure of what specific type of myotonic dystrophy they have (identified as unknown/unsure (U/U) throughout this report). These respondents are an important part of this study and are included in the **Overall** group. Instances where their experiences are significantly different are highlighted accordingly.

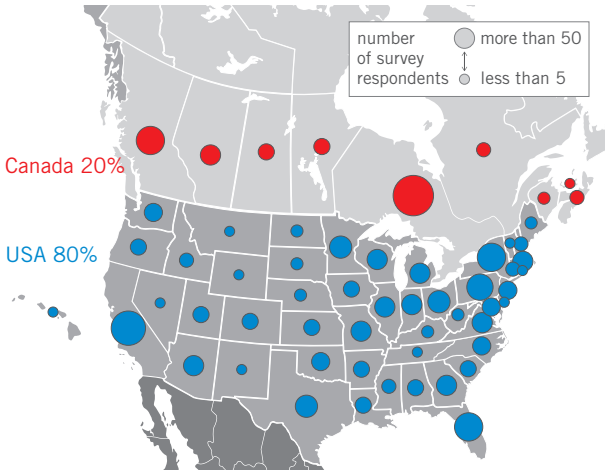
Patient Survey Respondents

The Christopher Project recruited participants from advocacy group mailing lists to capture input from a large and diverse group of respondents.

Geographic Location

Most Christopher Project respondents lived in the United States (80%), while 20% lived in Canada.

Generally, the reported experiences of American and Canadian respondents were similar.

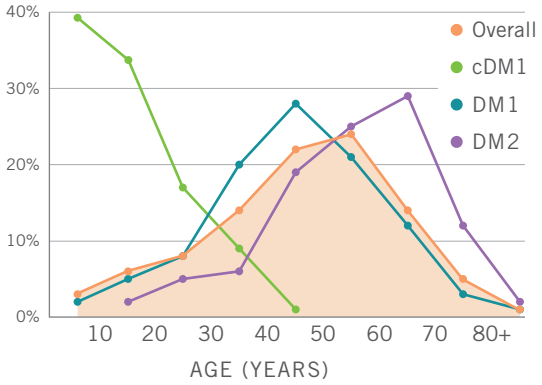


Age of Respondents

The results include feedback from respondents ranging in age from newborn to 86 years old. The average age **Overall** was 45 years old.

cDM1 respondents were much younger than all other respondents with an average age of 14.

DM2 respondents were slightly older with an average age of 55.

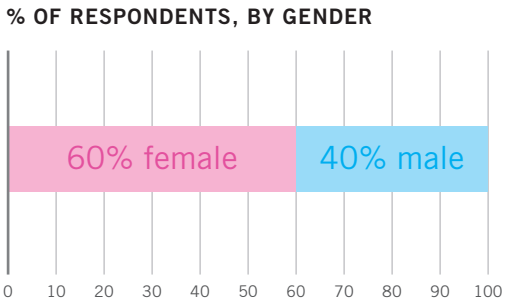


Gender

More women (60%) responded to the survey than men (40%).*

The survey findings for male and female respondents were very similar. Meaningful differences by gender were few and are highlighted in this report where appropriate.

*This represents the gender of survey respondents only. Myotonic dystrophy is equally prevalent across genders in the general population.

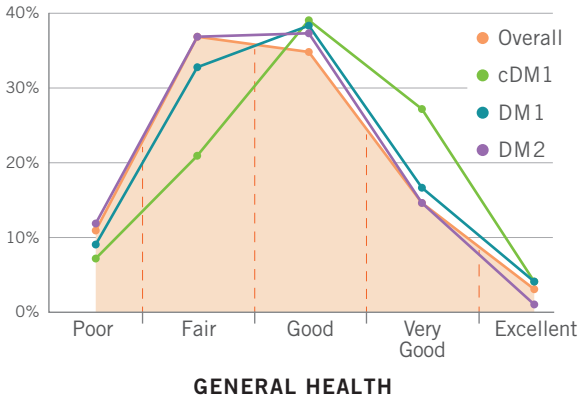


General Health

Approximately one half of **Overall** respondents indicated their health was excellent, very good, or good.

The other half of respondents indicated their health was generally fair or poor.

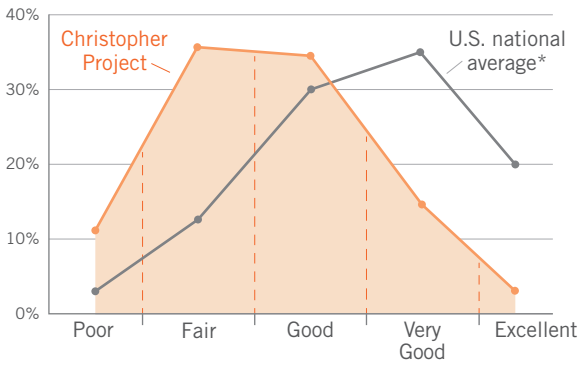
cDM1 respondents reported better general health than the other types.



GENERAL HEALTH

Comparative General Health

The general health reported by **Overall** respondents was relatively poor when compared to national averages (Canadian data not shown).



COMPARATIVE GENERAL HEALTH

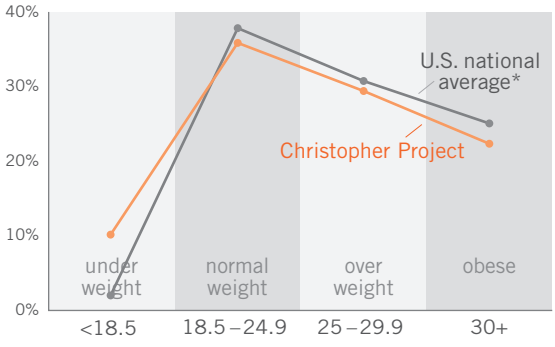
*Source: Maglinte et al, 2012

Body Mass Index

Body Mass Index (BMI) is a general health indicator, calculated as a ratio between weight and height (BMI=kg/m2).

The reported BMI of **Overall** adult respondents is consistent with national averages (Canadian data not shown).†

†Only adult respondents were included in this analysis.



BODY MASS INDEX (BMI)

*Source: NCCDPHP, 2015

Assistance with the Survey

The large majority of respondents were adults completing the survey on their own behalf. 11% of surveys were completed by a family member or caregiver responding on behalf of a patient under the age of 18.

39% of **Overall** respondents indicated that they had received at least some assistance from a spouse or parent/guardian in completing the survey. Most **cDM1** respondents (86%) reported major assistance, while **DM1** and **DM2** reported receiving minor or no assistance.

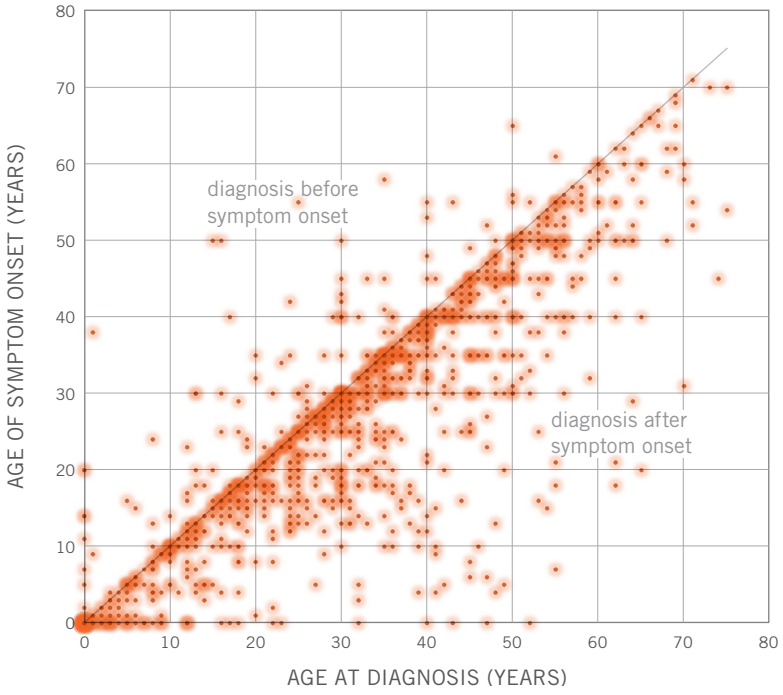
Receiving a Diagnosis

Receiving a diagnosis of myotonic dystrophy can be challenging—it can take years to get and, once received, can significantly impact the lives of patients and their families. Participants were asked a series of questions to get an understanding of their diagnostic experience.

Age of Onset and Age at Diagnosis

Respondents reported the age they experienced their first significant symptoms ***(age of symptom onset)*** and the age they received their medical diagnosis for myotonic dystrophy ***(age at diagnosis)***. These results are plotted on the chart below for **Overall** survey respondents. Each respondent is represented by a dot.

AGE OF SYMPTOM ONSET VS AGE AT DIAGNOSIS (OVERALL)



As shown, the age of onset and age at diagnosis varied greatly. In a limited number of cases, respondents received their diagnosis before onset of symptoms (top left), presumably because someone else in their family was diagnosed with this genetic condition before them.

In most cases, respondents received their diagnosis some time after onset of symptoms (bottom right). This time delay can be significant and is sometimes referred to as the ***diagnostic odyssey*** (Hilbert et al, 2013). These findings, **Overall** and by type, are summarized below.

	Overall	cDM1	DM1	DM2
Average age of onset	25.7	birth	27.2	36.5
Average age at diagnosis	30.1	0.2	31.5	43.7

	Overall	cDM1	DM1	DM2
Average diagnostic odyssey*	5.6	0.2	5.7	7.9

*calculated for respondents who reported diagnosis at or after symptom onset.

First Symptoms

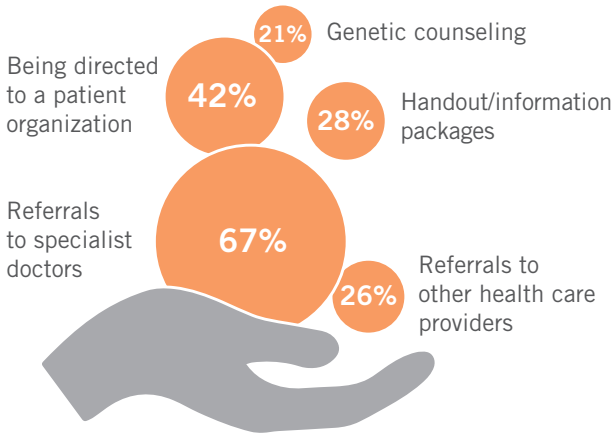
Overall	cDM1	DM1	DM2
Muscle weakness 41%	Muscle weakness 39%	Myotonia 35%	Muscle weakness 47%
Myotonia 28%	Neonatal issues 28%	Muscle weakness 35%	Myotonia 27%

Respondents consistently reported muscle weakness and myotonia as the first significant symptoms they noticed. The **cDM1** respondents also reported neonatal issues such as severe muscle weakness (hypotonia), respiratory complications, and feeding problems at birth.

Assistance Received at Diagnosis

Respondents were asked about the types of assistance they were given at the time of their diagnosis to gain an understanding of the support they received and what was most helpful.

TOP 5 TYPES OF ASSISTANCE RECEIVED



The five most common types of assistance given at the time of diagnosis (shown above) were also reportedly the **most helpful** when provided.

The types and level of assistance given to **DM1** and **DM2** respondents were similar. The **cDM1** respondents reported the most comprehensive support: referrals to other healthcare providers and school accommodations were provided to **cDM1** respondents **2.5 times** more often.

Other important findings around the experience of receiving a diagnosis include:

- **21%** of **Overall** respondents reported they were given **NO ASSISTANCE** at diagnosis.
- **Only 6%** reported receiving any psychological or emotional support, yet **52%** of respondents reported that the 'emotional impact' of receiving a diagnosis was a challenge.
- **43%** indicated that a 'lack of resources available about my disease' was a challenge.
- **Less than 5%** reported they were provided with any workplace accommodations.

Years Living with Myotonic Dystrophy

Because of its progressive nature, knowing how long respondents have lived with myotonic dystrophy adds context to their reported experience.

Overall	cDM1	DM1	DM2	U/U
15 yrs	14 yrs	13 yrs	12 yrs	19 yrs

For **Overall** respondents, it had been 15 years, on average, since they received their medical diagnosis for myotonic dystrophy. That said, results varied widely. Some survey respondents were newly diagnosed and had been living with myotonic dystrophy for a short time, others had been living with the disease for most of their adult lives. Notably, respondents who did not know or were unsure of their specific type (**U/U**) had been living with it significantly longer (19 years, on average).

Genetic Testing

Advances in medical genetics have made it possible to more accurately confirm a diagnosis of myotonic dystrophy.

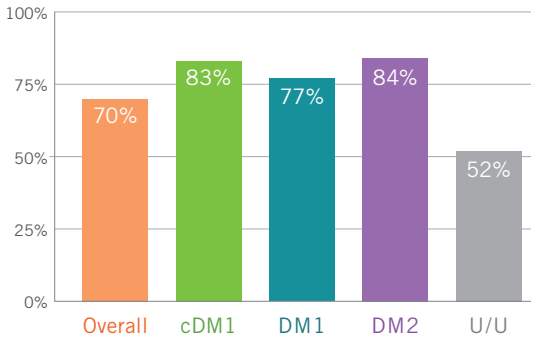
More than two-thirds of **Overall** respondents report that they have had their diagnosis confirmed through genetic testing.

Some reasons offered for not receiving genetic testing include:

- Others in family had already received genetic confirmation
- Clinical presentation alone was sufficient for diagnosis
- Genetic testing not offered/available*
- Alternative testing used (muscle biopsy or electromyography)
- Cost and/or insurance risk too high

*many respondents were diagnosed before genetic testing became available (around 1992 for **DM1** and 2001 for **DM2**)

% WITH GENETIC CONFIRMATION

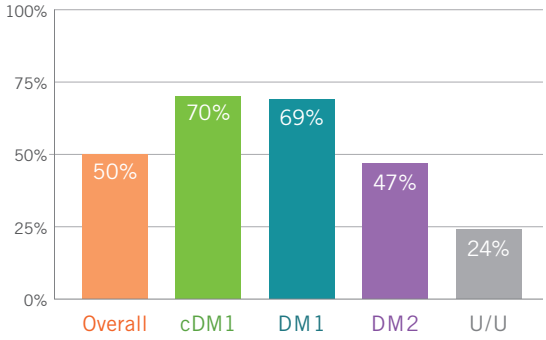


Repeat Count

Some people are provided with a repeat count or repeat number at the time of their genetic confirmation.

50% of **Overall** respondents indicated they received a repeat count, as shown. Significantly fewer **DM2** respondents reported they received a repeat count compared to **cDM1** and **DM1** respondents.

% WHO RECEIVED REPEAT COUNT

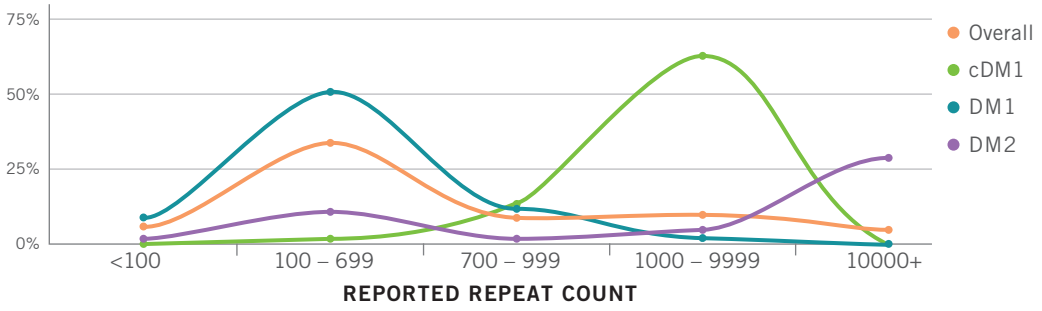


'Repeat count' is a term used to describe the nature of the mutations that cause the different types of myotonic dystrophy.

- The typical repeat count for **DM1** is greater than 50 and the typical repeat count for **cDM1** is greater than 1,000 (Bird, 1999).
- For **DM2**, the typical repeat count range is 75 to 10,000 with an average of about 5,000 (Dalton, Ranum & Day, 2006).

Repeat count numbers have not been directly correlated to symptom severity, they may vary between different tissues throughout the body, and they can change over time.

The repeat count numbers for survey respondents who provided this information are illustrated below.



The reported repeat counts generally follow the typical patterns seen in the different types of myotonic dystrophy, as described above. For more details on the genetics of myotonic dystrophy visit the following links:

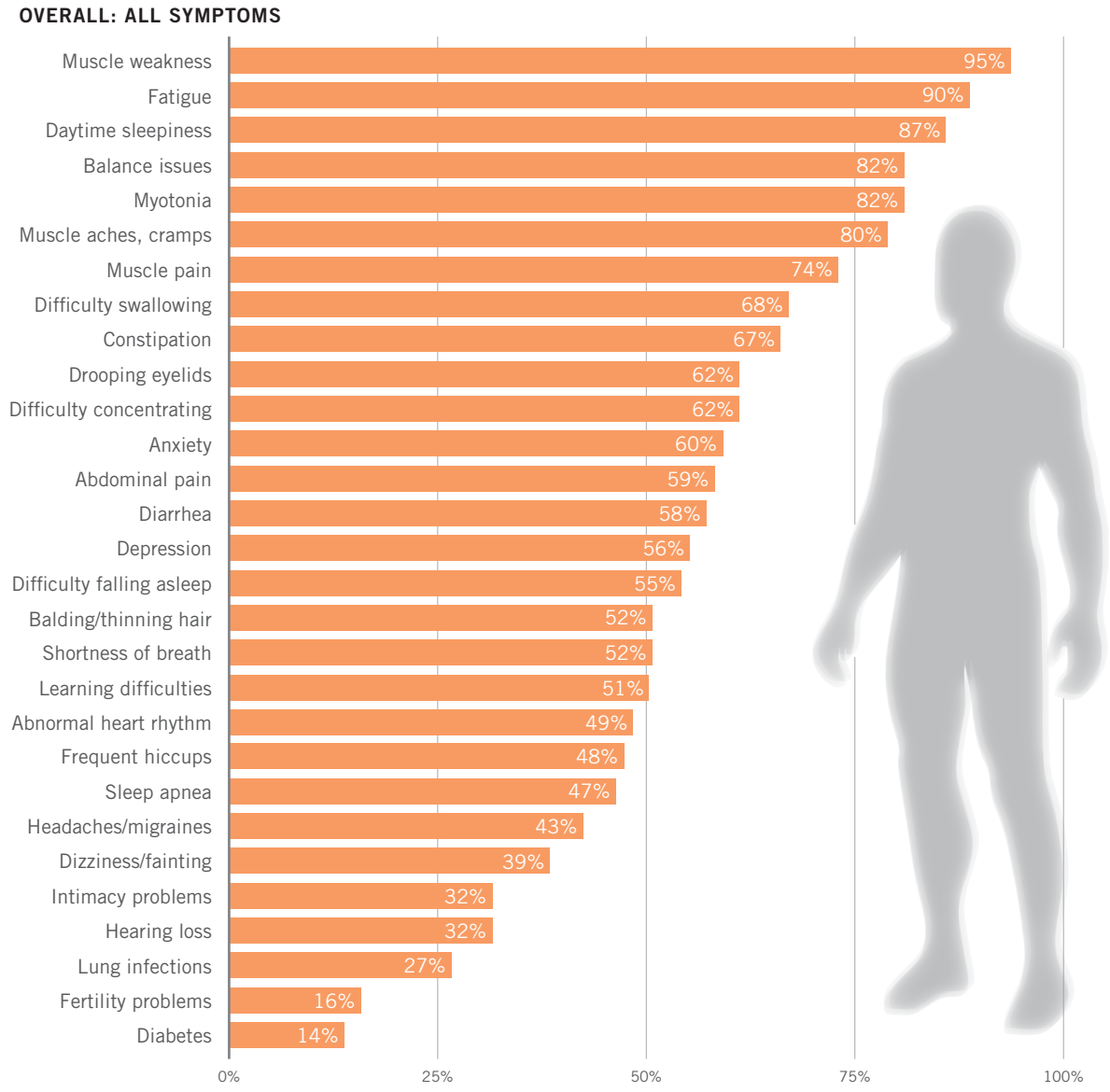
- www.mda.org/disease/myotonic-dystrophy/causes-inheritance (MDA, 2018)
- www.myotonic.org/causes-myotonic-dystrophy-0 (MDF, 2018)

Symptoms

Myotonic dystrophy affects many different parts of the body and not everyone experiences the same symptoms in the same way. Respondents were asked to rate 29 symptoms across seven categories in order to understand how many people experienced the symptom (prevalence) and its impact on daily life.

Symptom Prevalence

The percentage of respondents who reported that they experience a particular symptom is shown below:

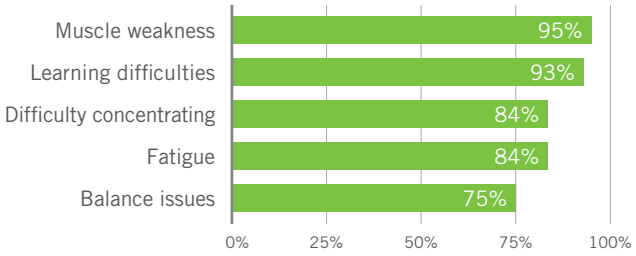


Muscle weakness, fatigue, and daytime sleepiness were the most prevalent symptoms.

Symptom Prevalence by Type

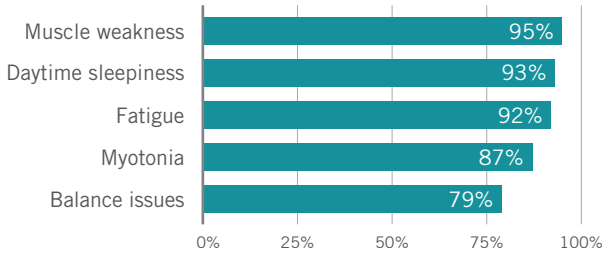
There was some variation in the most prevalent symptoms among the different types of myotonic dystrophy (below). All respondents, regardless of type, reported muscle weakness as the most prevalent symptom.

cDM1: TOP 5



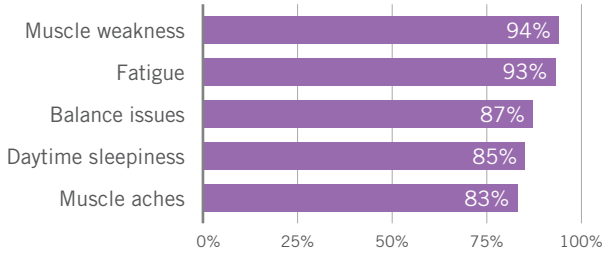
- Learning difficulties and difficulty concentrating were more prevalent in the **cDM1** group compared to other groups.

DM1: TOP 5



- Daytime sleepiness and myotonia were more prevalent in the **DM1** group compared to other groups.

DM2: TOP 5



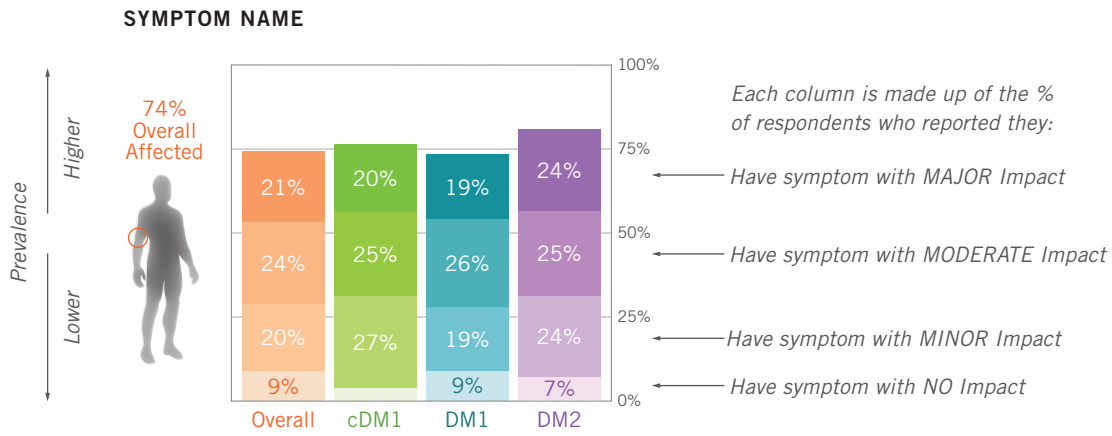
- Balance issues and muscle aches/cramps were more prevalent in the **DM2** group compared to other groups.

Symptom Impact

The level of impact each symptom or physical problem had on respondents' daily lives is presented graphically on the following pages, broken down by symptom category:

- Muscle Symptoms
- Gastrointestinal Symptoms
- Cardiorespiratory (Heart and Lung) Symptoms
- Sleep and Fatigue Symptoms
- Psychological (Cognitive) Symptoms
- Hormonal and Endocrine Symptoms
- Other Symptoms

The key below explains how to interpret the graphic results:



- *Higher percentages (taller bars) mean many people experience this symptom*
- *Lower percentages (shorter bars) mean fewer people experience it*
- *Colored bars reflect responses for each group*
- *The darker the color the more significant the symptom impact*
- *The **Overall Affected** is the total % of all respondents who experience the symptom (prevalence)*

In the above example:

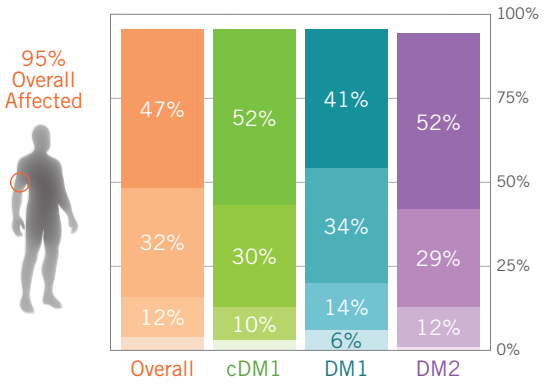
- 74% of **Overall** respondents are reportedly affected by this particular symptom.
- The **DM2** group is reportedly more affected by this particular symptom (highest bar), with 24% having reported 'MAJOR' impact on their daily life.

Muscle Symptoms

Muscle-related symptoms were the most common complaint for survey respondents:

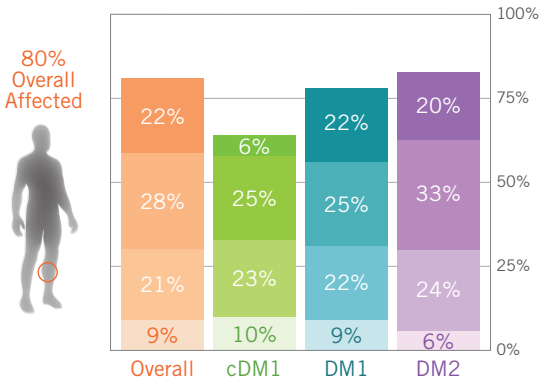
- They accounted for four of the seven most prevalent symptoms overall
- Their impact on daily life was substantial
- They play an integral role in many other symptoms

MUSCLE WEAKNESS



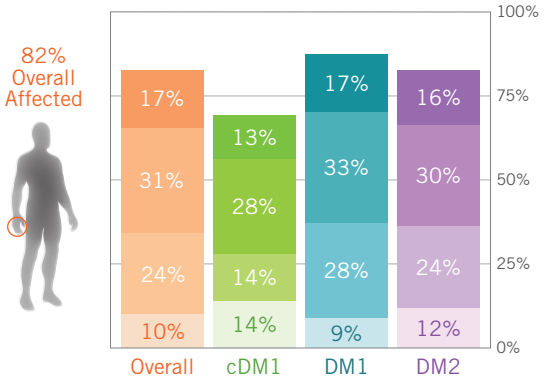
Muscle weakness was the most prevalent and impactful symptom: 95% of **Overall** respondents experienced it and 47% reported it has a major impact on daily life.

MUSCLE ACHES, CRAMPS



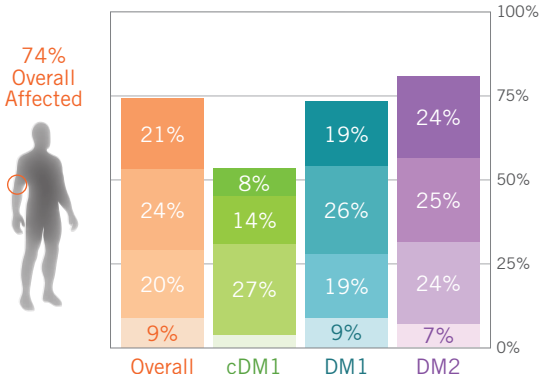
80% of **Overall** respondents experienced muscle aches and cramps and **DM2** respondents reported the highest level of impact.

MYOTONIA (DIFFICULTY RELAXING MUSCLES)



While a large majority (82%) of **Overall** respondents experienced myotonia, the relative proportion reporting major impact from myotonia was less than other muscle symptoms.

MUSCLE PAIN



Three quarters of **Overall** respondents experienced muscle pain. **DM2** respondents experienced more muscle pain with more impact.

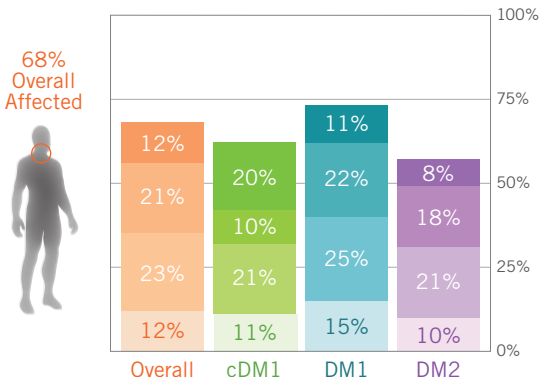
Muscle complaints were reportedly less common and less impactful for the **cDM1** group, except muscle weakness, which affected all groups equally.

Gastrointestinal Symptoms

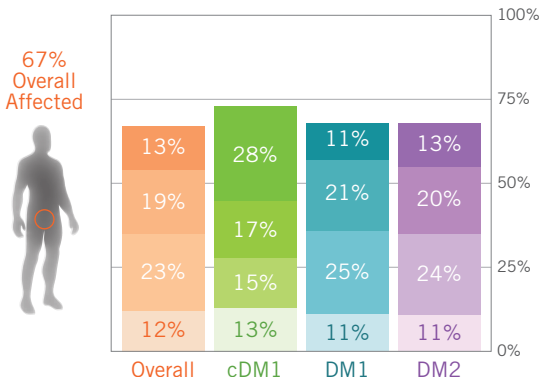
Gastrointestinal (GI) symptoms presented significant health issues for Christopher Project respondents:

- 91% of respondents reported they experience at least one GI symptom
- 64% reported that one or more GI symptom(s) had a significant (major/moderate) impact on daily life

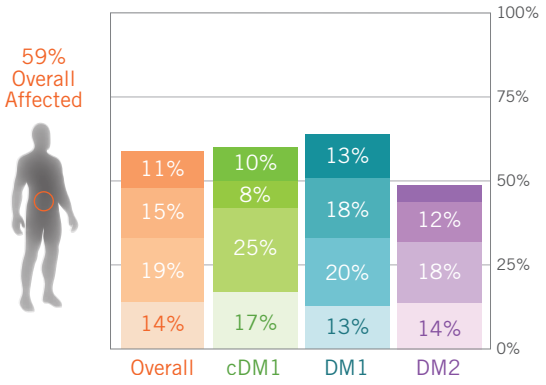
DIFFICULTY SWALLOWING



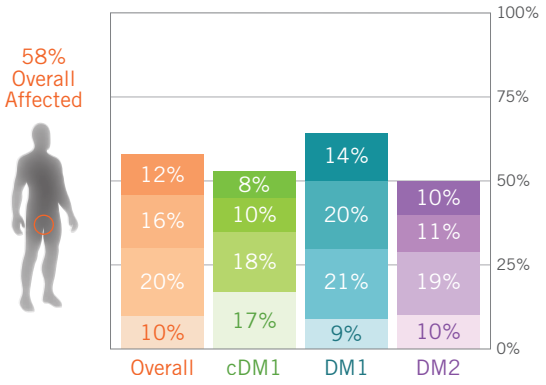
CONSTIPATION



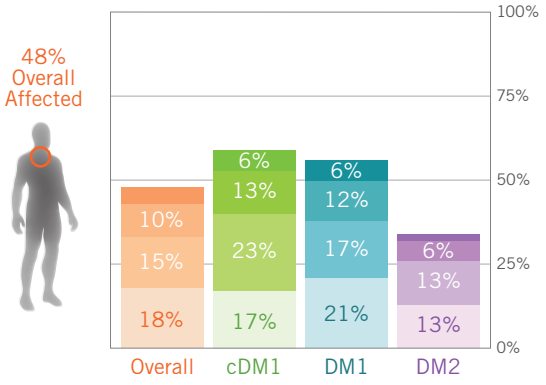
ABDOMINAL PAIN



DIARRHEA



FREQUENT HICCUPS



DM1 respondents reported the highest impact due to difficulty swallowing, abdominal pain, and diarrhea.

cDM1 respondents reported they were more impacted by frequent hiccups and constipation.

DM2 respondents experienced gastrointestinal symptoms less frequently and with less impact, except constipation.

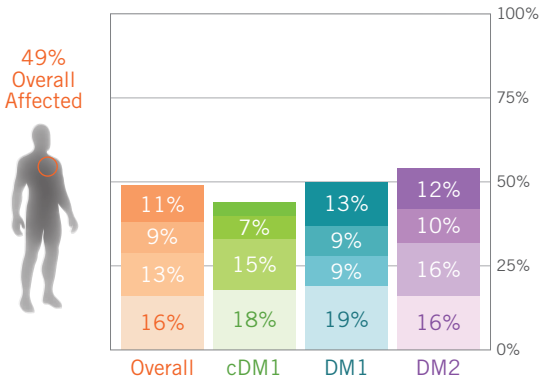
Significantly more females than males reported major or moderate impact from abdominal pain, diarrhea, and constipation (data by gender not shown).

Cardiorespiratory (Heart and Lung) Symptoms

Overall, cardiorespiratory (heart and lung) symptoms were less prevalent and reportedly less impactful than muscle symptoms. However, they pose a significant life risk for those who experience them:

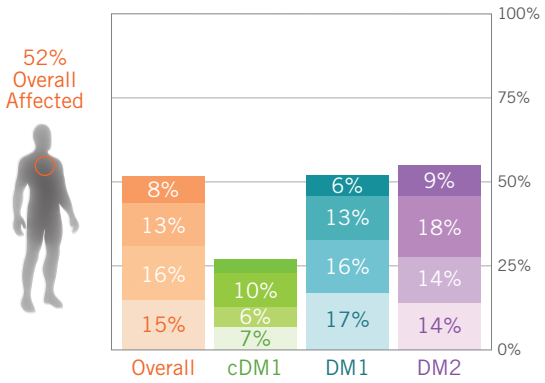
- 77% of respondents reported they are impacted by at least one cardiorespiratory symptom
- 39% indicated one or more of them had a significant (major/moderate) impact on daily life

ABNORMAL HEART RHYTHM



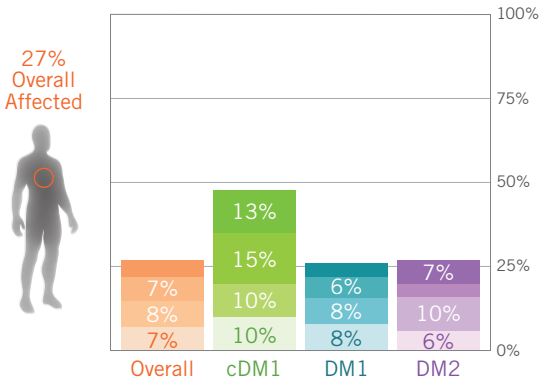
Half of **Overall** respondents reported having an abnormal heart rhythm. Additional findings on cardiac issues are in the *Cardiac Risk Awareness* section later in this report.

SHORTNESS OF BREATH



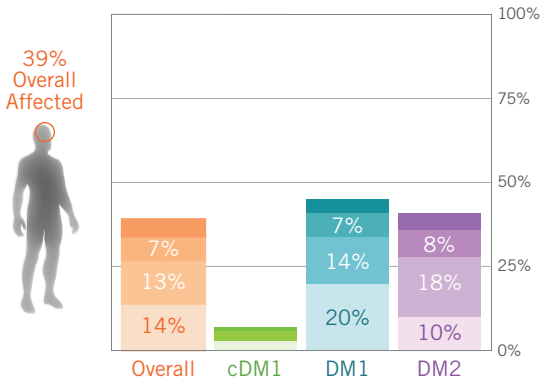
The **cDM1** group reported they were less affected by shortness of breath than both **DM1** and **DM2** respondents.

RECURRENT LUNG INFECTIONS (PNEUMONIA)



The **cDM1** group experienced lung infections twice as often as other respondents with greater impact on their daily lives (13% major, 15% moderate).

DIZZINESS / FAINTING



DM1 and **DM2** respondents were significantly more affected by dizziness/fainting than the **cDM1** group. Significantly more females than males reported major or moderate impact from dizziness/fainting (data by gender not shown).

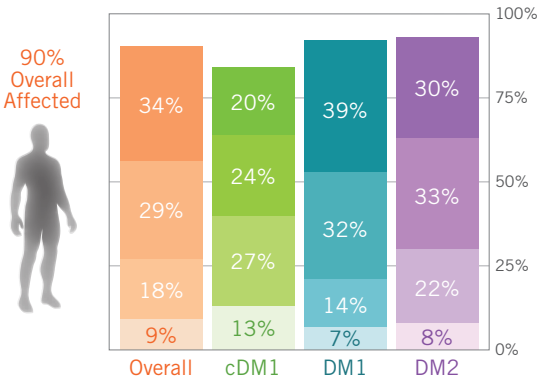
It is critical that patients who experience any cardiorespiratory symptoms speak with a healthcare provider about the associated risks and receive appropriate medical care and advice accordingly.

Sleep and Fatigue Symptoms

Sleep and fatigue issues can be pervasive and present significant challenges in daily life:

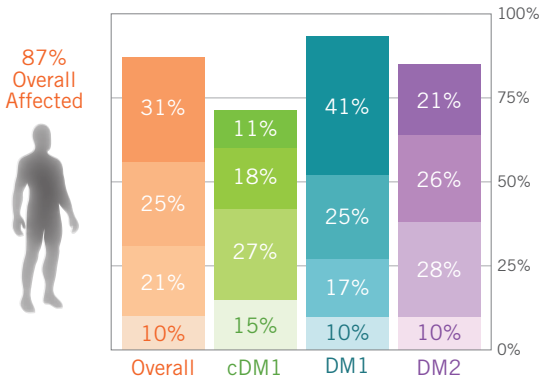
- 29% of respondents indicated they experienced ALL of these symptoms
- 95% reported they experience at least one of them
- 74% indicated one or more of them had a significant (major/moderate) impact on daily life

FATIGUE



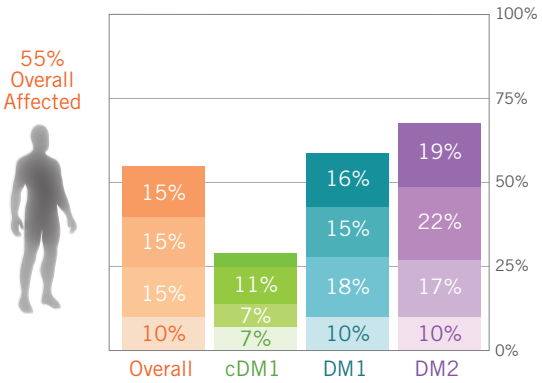
Fatigue was the second most prevalent and impactful symptom **Overall**, affecting 90% of respondents. More **DM1** respondents reported major impact from fatigue than any other group.

DAYTIME SLEEPINESS



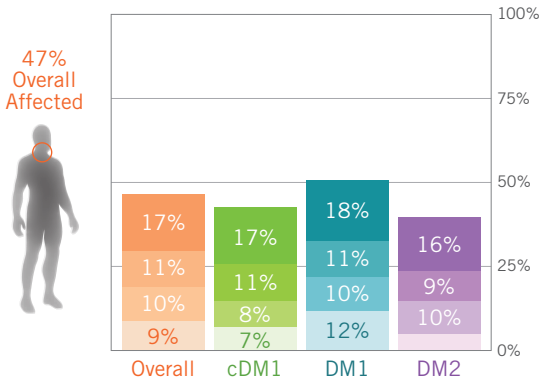
Daytime sleepiness was the third most prevalent and impactful symptom, affecting 87% of respondents. The **DM1** group reported the highest level of impact from this symptom.

DIFFICULTY FALLING ASLEEP



55% of **Overall** respondents experienced difficulty falling asleep, with **DM2** respondents experiencing greater difficulty falling asleep than other groups.

TROUBLE BREATHING DURING SLEEP (APNEA)



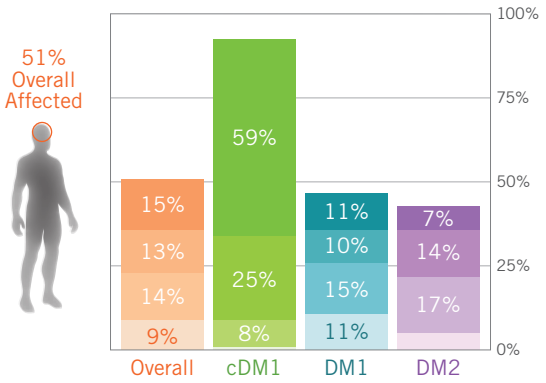
47% of all respondents reported they experienced sleep apnea.

Psychological (Cognitive) Symptoms

Psychological symptoms and related cognitive issues were common and presented significant challenges:

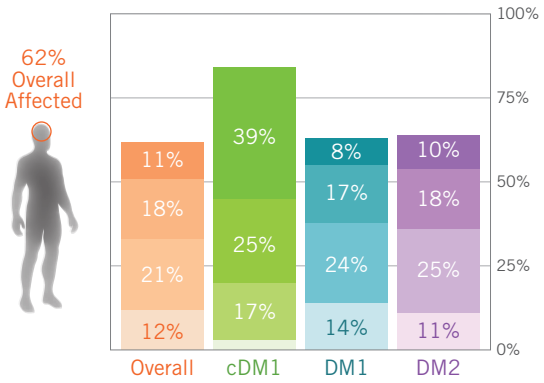
- 32% of respondents indicated that they were impacted by ALL of these symptoms
- 71% reported they were impacted by at least one of these symptoms
- 52% reported one or more of them had a significant (major/moderate) impact on daily life

LEARNING DIFFICULTIES



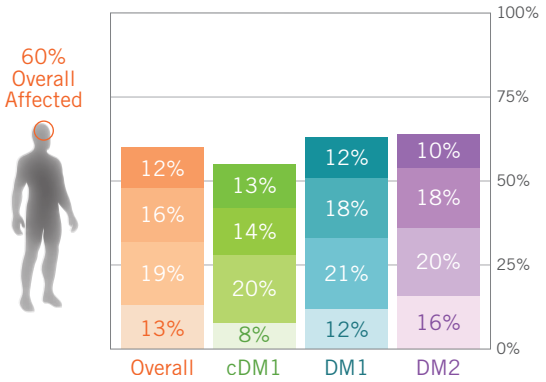
51% of **Overall** respondents reported having learning difficulties. 93% of **cDM1** respondents reported having them with significantly greater impact. Significantly more males than females reported they were impacted by learning difficulties (data by gender not shown).

DIFFICULTY CONCENTRATING



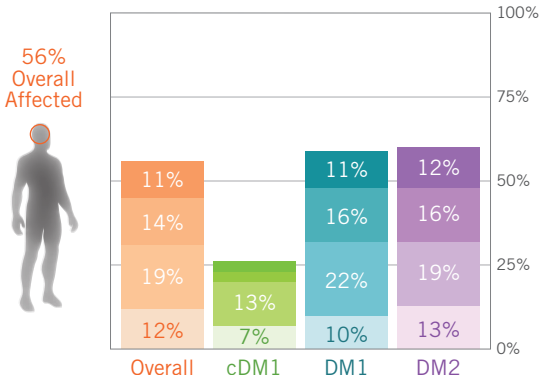
62% of **Overall** respondents reported difficulty concentrating. The **cDM1** group reported greater difficulty, with 84% experiencing difficulty concentrating (39% major impact).

ANXIETY



60% of **Overall** respondents reported they experienced at least some level of anxiety.

DEPRESSION



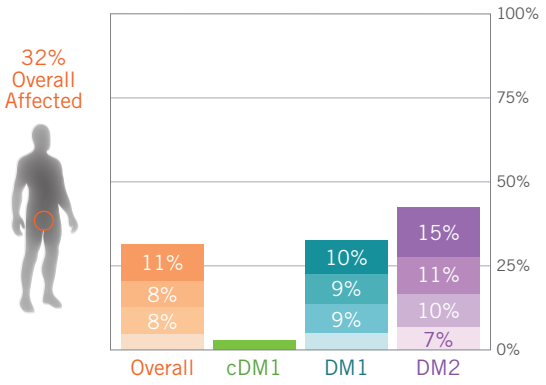
56% of **Overall** respondents reported they experienced at least some level of depression. This number was significantly lower for the **cDM1** group.

Hormonal and Endocrine Symptoms

Respondents experienced these issues less frequently than most other symptoms. However, for those who experienced them, the impact was relatively high:

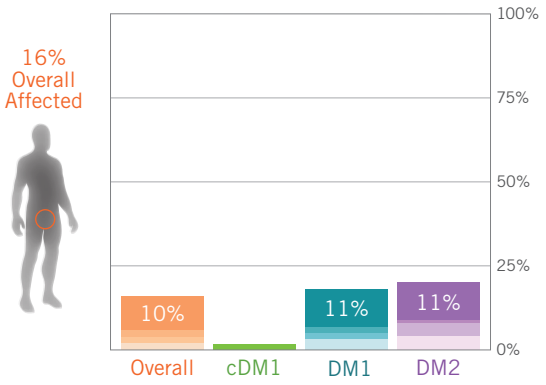
- The prevalence and impact of these symptoms varied depending on respondents' age, gender, and type of myotonic dystrophy

SEXUAL / INTIMACY PROBLEMS



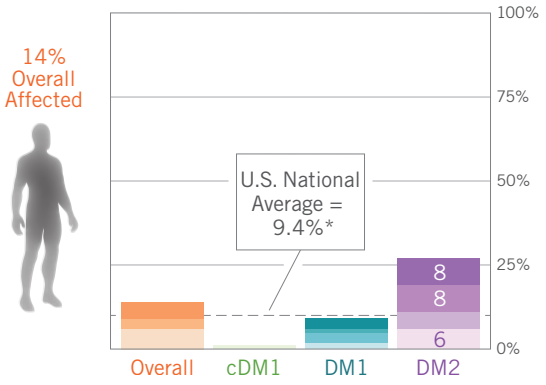
32% of **Overall** respondents reported they experienced sexual/intimacy problems. Significantly more males than females reported major or moderate impact from intimacy problems (data by gender not shown).

FERTILITY PROBLEMS



16% of **Overall** respondents reported they experienced fertility problems. Almost all respondents who had fertility problems reported major impact. 19% of female respondents reported fertility problems compared to 13% of male respondents (data by gender not shown).

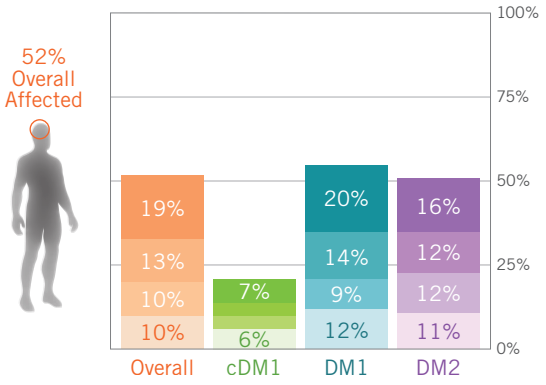
DIABETES



The reported prevalence of diabetes by the **DM2** group was significantly higher than the U.S. national average.

*% of US population with type 1 or type 2 diabetes; (CDC National Diabetes Statistics Report, 2017)

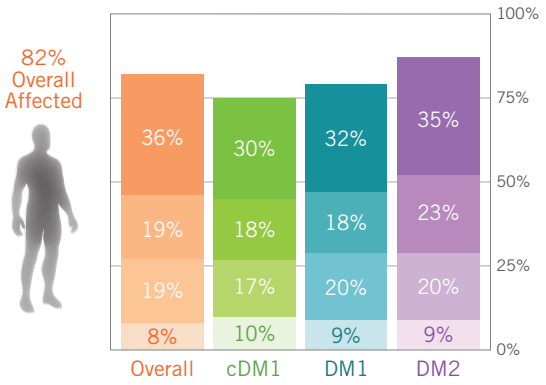
HAIR LOSS



Over half of **Overall** respondents (52%) reported they experienced hair loss/balding. Significantly more males (74%) than females (38%) reported they experienced hair loss (data by gender not shown).

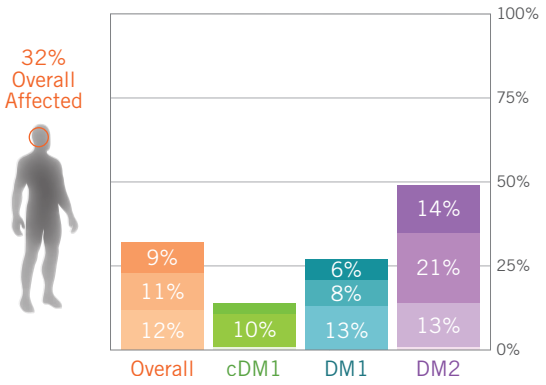
Other Symptoms

BALANCE ISSUES



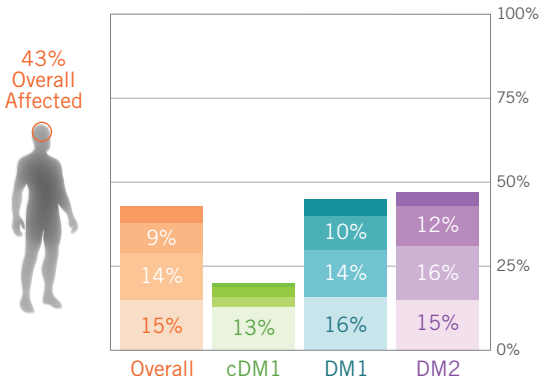
Balance issues were the fourth most prevalent symptom, with 82% of **Overall** respondents reporting they experienced them (36% major impact).

HEARING LOSS



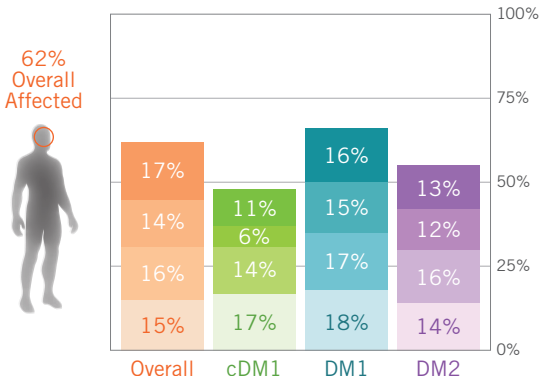
32% of **Overall** respondents reported they experienced some level of hearing loss. More **DM2** respondents had it and reported greater impact from this symptom (14% major, 21% moderate impact).

HEADACHES / MIGRAINES



43% of **Overall** respondents experienced headaches/ migraines. The **cDM1** group experienced these symptoms less than other groups. Significantly more females than males reported major or moderate impact from headaches/migraines (data by gender not shown).

DROOPING EYELIDS (PTOSIS)



62% of **Overall** respondents reported they had drooping eyelids, which can significantly affect vision. Drooping eyelids impacted **DM1** respondents more than other groups.

Managing Daily Life

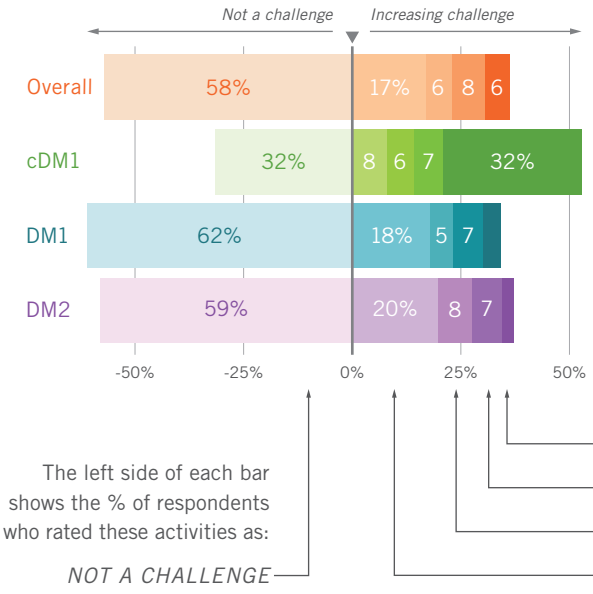
Given the diversity of symptoms and their progressive nature, daily activities can be challenging for some individuals living with myotonic dystrophy.

Challenges with Daily Life Activities

Survey respondents were asked to rate the level of challenge they experience in performing specific activities in daily life. Answers were grouped into eight major categories:

- Mobility Activities
- Household and Personal Needs Activities
- Psychological (Cognitive) Activities
- Communication Activities
- Social Activities
- Doctors Office Activities
- School Activities
- Work Activities

The results for each category are presented graphically on the following pages. The key below shows how to interpret the graphics:



- Results shown are for all activities included in the category (combined and averaged)
- Numbers reflect the % of respondents for each rating level (values less than 5% not shown)
- Colored bars reflect responses for each type group
- Lightest shade means not a challenge (left side)
- Darker shades mean more challenge (right side)

The left side of each bar shows the % of respondents who rated these activities as:

NOT A CHALLENGE

The right side of each bar shows the % of respondents who rated these activities as:

UNABLE to Perform Activity

a MAJOR Challenge

a MODERATE Challenge

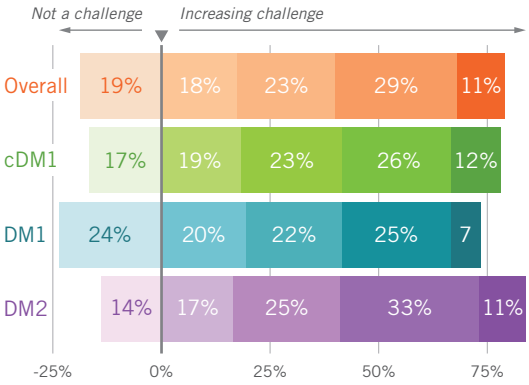
a MINOR Challenge

In the above example:

- 58% of **Overall** respondents reported activities in this category were 'NOT A CHALLENGE' to perform.
- Activities in this category were more challenging for the **cDM1** group (32% 'UNABLE to perform').

Mobility Activities

This category includes: going up and down stairs, standing, maintaining balance, standing up/sitting down/bending down, walking

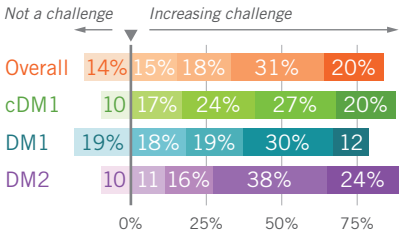


Mobility activities were the most challenging category of daily life activities:

- 81% of **Overall** respondents experienced some level of challenge with mobility (29% major challenge; 11% unable to perform).
- **DM2** respondents experienced greater challenges with mobility.

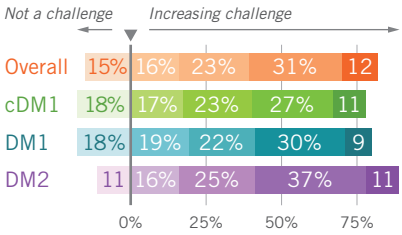
A closer look at the most challenging Mobility Activities:

GOING UP AND DOWN STAIRS



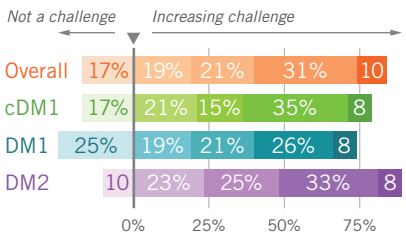
- Going up and down stairs was the most challenging mobility activity **Overall** (31% major challenge; 20% unable to perform).
- **DM2** respondents were particularly challenged going up and down stairs (38% major challenge; 24% unable to perform).

STANDING (FOR ANY LENGTH OF TIME)



- Standing was the second most challenging mobility activity **Overall** (31% major challenge; 12% unable to perform).
- Standing was more challenging for **DM2** respondents (37% major challenge; 11% unable to perform).

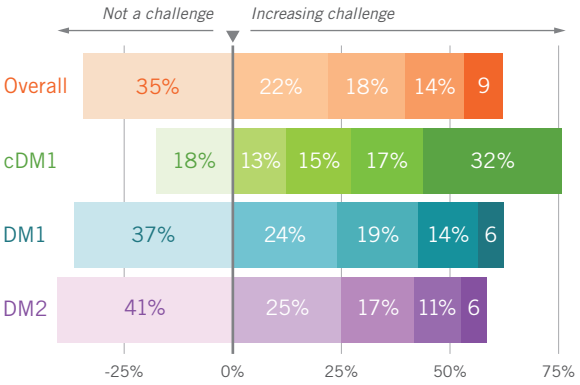
MAINTAINING BALANCE



- Maintaining balance was the third most challenging mobility activity **Overall** (31% major challenge; 10% unable to perform).
- Maintaining balance was more challenging for **DM2** respondents.

Household and Personal Needs Activities

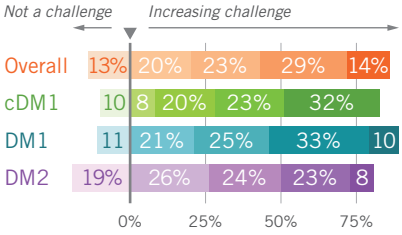
This category includes: handling objects, housekeeping, preparing meals, dressing, washing, using cutlery, swallowing (eating/drinking)



- 63% of **Overall** respondents reported some level of challenge associated with household duties and personal needs activities.
- The **cDM1** group reported a greater level of challenge in performing all household and personal needs activities than any other group.
- Significantly more males than females reported greater challenges with dressing, washing, and swallowing (data by gender not shown).

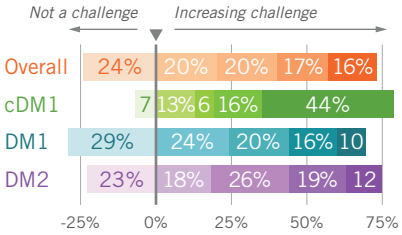
A closer look at the most challenging Household and Personal Needs Activities:

HANDLING OBJECTS



- Handling objects was one of the most challenging activities for **Overall** respondents across all categories (29% major challenge; 14% unable to perform).
- The **cDM1** group reported the greatest challenge handling objects (23% major challenge; 32% unable to perform).
- **DM1** respondents reported significant difficulty handling objects (33% major challenge; 10% unable to perform).

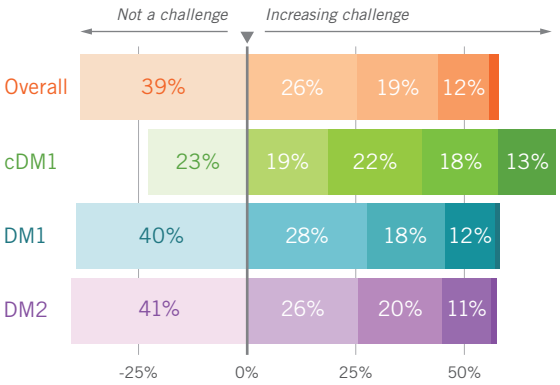
HOUSEKEEPING



- Housekeeping was the second most challenging household activity for **Overall** respondents (17% major challenge; 16% unable to perform).
- Housekeeping was most challenging for **DM2** and **cDM1** respondents.

Psychological (Cognitive) Activities

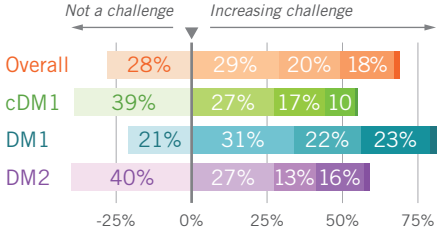
This category includes: staying alert, remembering things, concentrating, putting thoughts into words, planning daily activities



- 59% of **Overall** respondents indicated psychological activities present a challenge in their daily lives.
- While the severity of challenge reported for psychological activities is lower than other categories, the **Overall** number of respondents expressing at least some level of challenge in this area was relatively high.

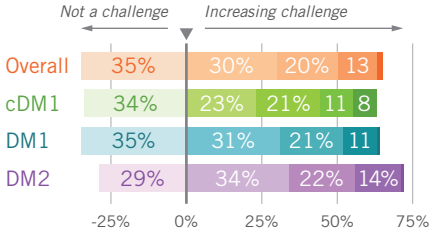
A closer look at the most challenging Psychological (Cognitive) Activities:

STAYING ALERT



- Staying alert was the most challenging psychological activity **Overall** (18% major challenge; 2% unable to stay alert)
- **DM1** respondents reported the greatest challenge with staying alert (23% major challenge; 2% unable to perform)

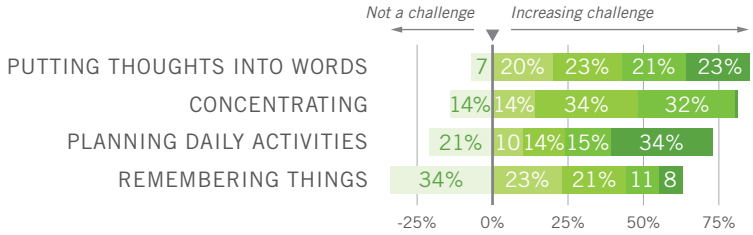
REMEMBERING THINGS



- Remembering things was the second most challenging psychological activity **Overall** (13% major challenge; 2% unable to perform)
- **DM2** respondents reported greater difficulty remembering things than the **DM1** group (15% major challenge; 1% unable to perform)

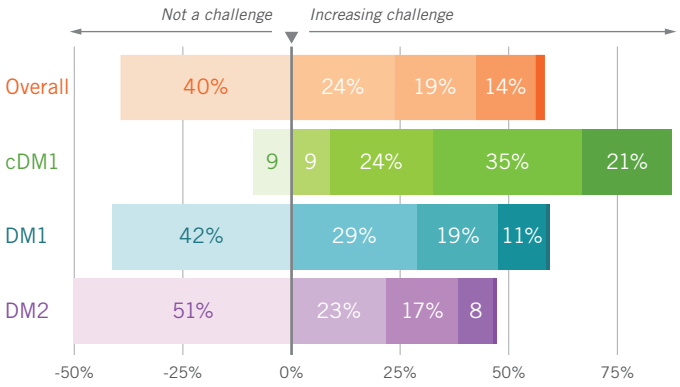
A closer look at the **cDM1** experience with Psychological (Cognitive) Activities:

When compared to other respondents, the **cDM1** group reported substantially greater challenges in all psychological activities except staying alert.



Communication Activities

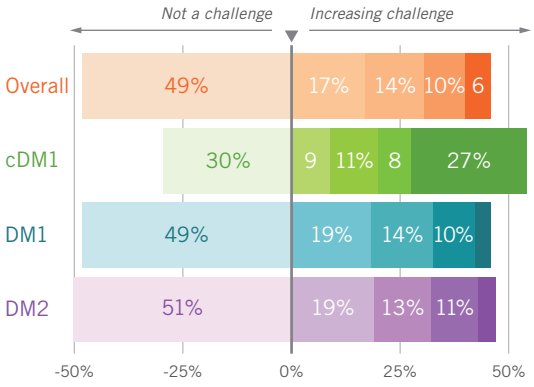
This category includes: speaking (pronouncing words), writing (holding a pen)



- 60% of **Overall** respondents experienced at least some level of challenge with communications activities.
- 89% of **cDM1** respondents experienced at least some level of challenge with both writing and speaking with 35% reporting major challenge and 21% reporting they are unable to perform these activities.
- Significantly more males than females reported greater challenges with speaking and writing (data by gender not shown).

Social Activities

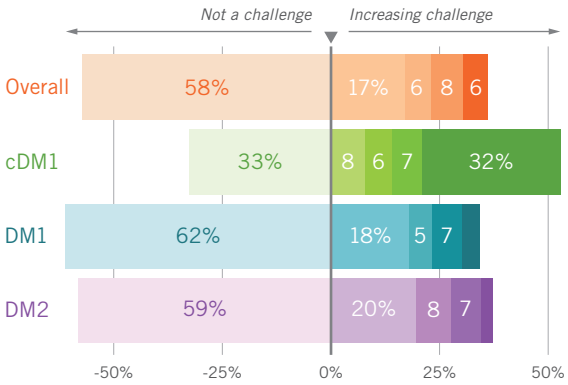
This category includes: disclosure (talking about my disease), relationships/interactions with others, romantic/emotional/intimate life



- 47% of **Overall** respondents reported at least some level of challenge with social activities with 10% indicating social activities were a major challenge for them and 6% reporting that they were unable to perform these activities.
- The **cDM1** group reported greater social challenges than other groups, particularly with disclosure (talking about their disease).
- Significantly more males than females reported greater challenges with romantic/emotional/intimate life and disclosure (data by gender not shown).

Doctors Office Activities

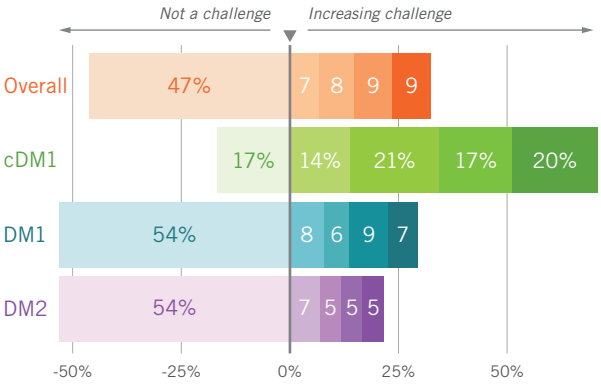
This category includes: preparing for a visit to the doctor, advocating for appropriate care, booking/tracking appointments



- 58% of **Overall** respondents indicated Doctors Office Activities did not present a challenge for them yet 37% reported at least some level of challenge.
- The **cDM1** group reported substantially greater challenges in this area (32% are unable to perform these activities).

School Activities

This category includes: completing education/schooling



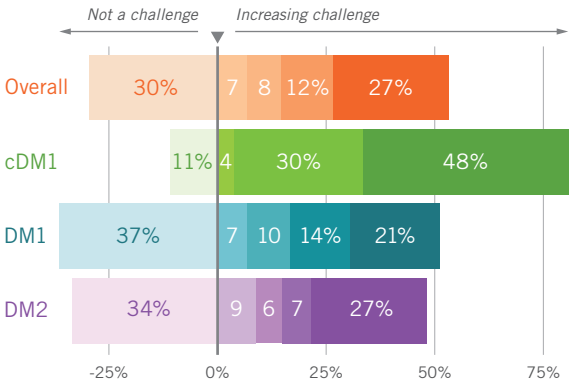
Note: Results shown are for all respondents, including those attending school. Only 80% of respondents provided a rating for this particular item.

- 47% of **Overall** respondents indicated it was <u>not</u> a challenge to complete their education/schooling.
- However, 33% reported at least some difficulty in doing so, with 9% reporting major challenges and a further 9% reporting that it was not possible to complete their education.
- **cDM1** respondents reported considerably more challenges with school (17% major challenge; 20% unable to complete their education/schooling).
- Significantly more males than females reported greater challenges with completing education/ schooling (data by gender not shown).

For additional information on education attainment see the *More About Daily Life* section.

Work Activities

This category includes: employment (obtaining/retaining work)



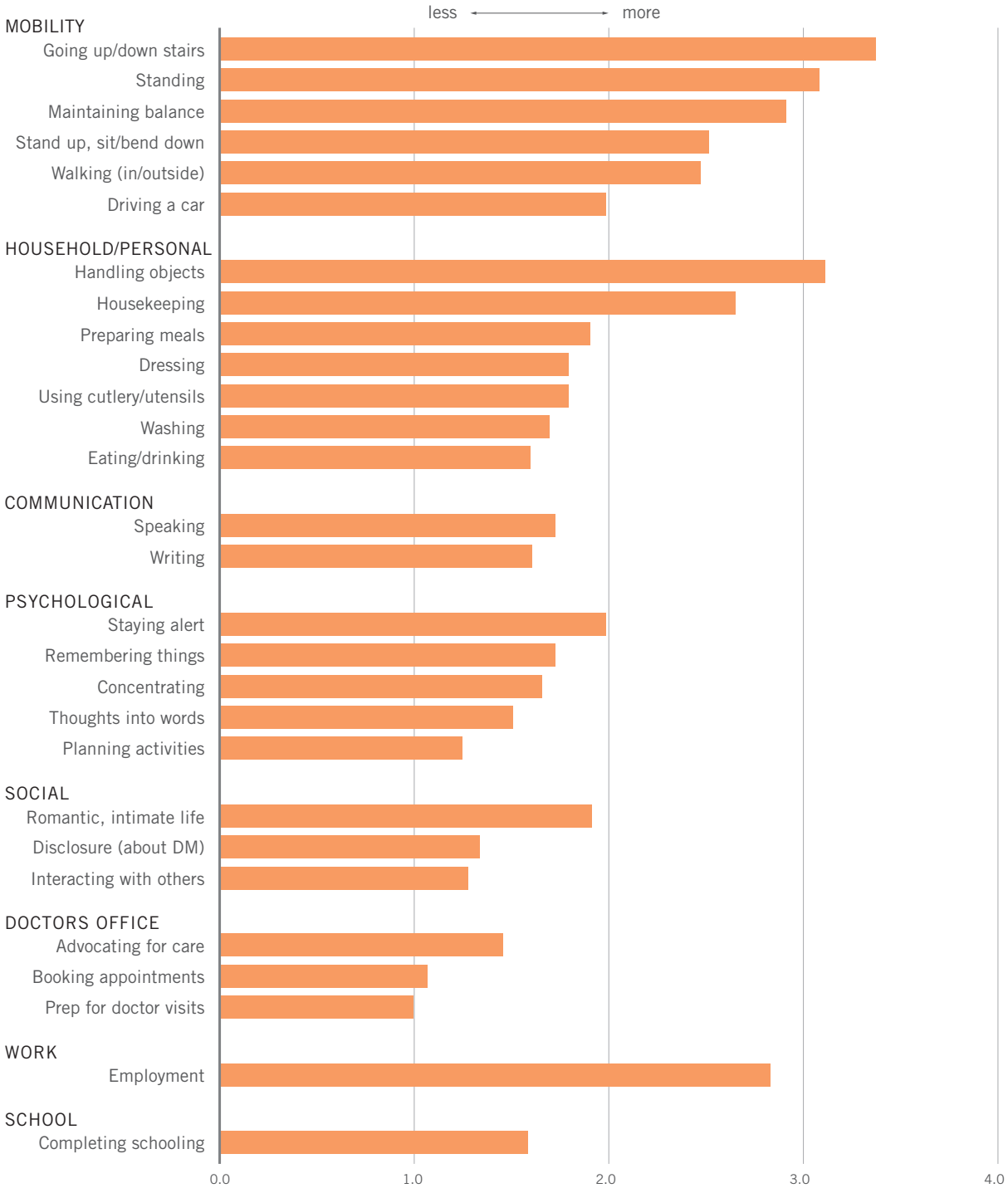
Note: Results shown are for respondents aged 16 to 64 (n=945).

- 54% of **Overall** respondents aged 16 to 64 indicated at least some level of challenge with employment (obtaining/retaining work), with 27% reporting they were unable to work at all.
- **cDM1** respondents aged 16 to 64 reported the greatest level of challenge associated with employment, with 48% reporting they were unable to work at all.
- Significantly more males than females reported greater challenges with employment (data by gender not shown).

Further details on employment are presented in the *More About Daily Life* section.

Relative Challenge of All Daily Life Activities

The chart below shows the relative challenge for **Overall** respondents for each of the individual daily life activities.



NOTE: Results shown are weighted by the reported level of challenge for each activity (MINOR=1.0, MODERATE=2.0, MAJOR=3.0, and UNABLE TO PERFORM=4.0) using valid responses only (missing values excluded).

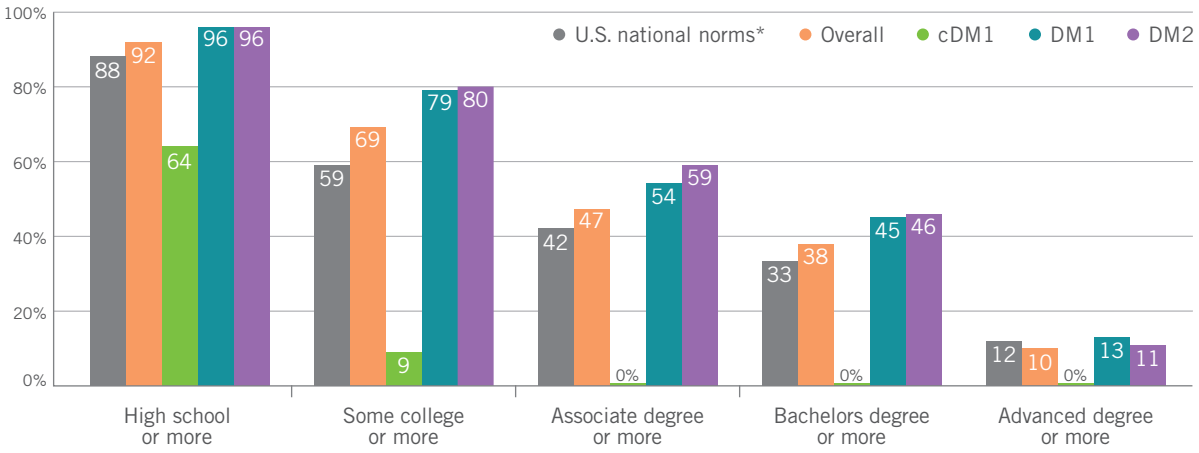
As shown, the five most challenging activities were going up and down stairs, handling objects, standing (for any length of time), maintaining balance, and employment.

More About Daily Life

Education, employment, income, and living situation play an important role in quality of life and have implications for social participation, self-esteem, and financial security.

Education Attainment for Respondents Aged 25 and Older

The highest level of education a person has achieved is known as *education attainment*.



All numbers in percent (%)

*Source: U.S. Census Bureau, 2015(a)

The education attainment reported by **Overall** respondents aged 25 and older generally aligned with the U.S. national average for the same age group. Canadian national norms are similar (data not shown).

The education attainment reported by **DM1** and **DM2** groups was significantly higher than U.S. and Canadian national averages (Canadian data not shown).

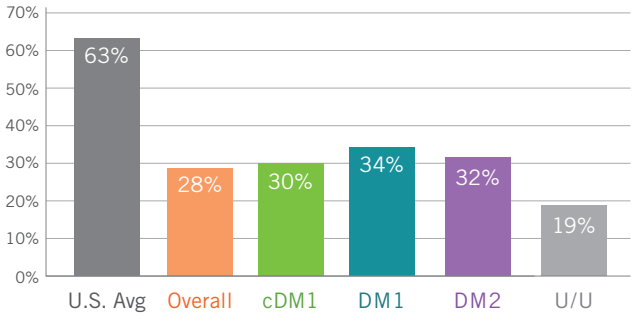
The majority of **cMD1** respondents were under the age of 25 and still in school (55%) or of preschool age (13%). As such, the majority of cDM1 respondents are not included in the above chart.

Labor Force Participation

The *labor force participation rate* is defined as the percentage of people aged 16 to 64 who are employed (full-time or part-time) or actively looking for work.

The labor force participation rate of **Overall** Christopher Project respondents was 28%, significantly lower than the U.S. average of 63% and the Canadian average of 65% (U.S. Census Bureau, 2015(b); Statistics Canada, 2016(a)).

LABOR FORCE PARTICIPATION RATE



Employment

Survey respondents aged 16 to 64 provided the following information on their current employment status:

	Overall (n=945)	cDM1 (n=27)	DM1 (n=400)	DM2 (n=138)	U/U (n=287)
Student	6%	22%	7%	3%	4%
Employed full-time	14%	4%	17%	22%	7%
Employed part-time	11%	19%	13%	8%	10%
Retired	11%	0%	10%	17%	11%
Unemployed by choice	8%	0%	13%	4%	5%
Seeking employment	3%	7%	4%	2%	2%
Unable to work due to myotonic dystrophy	52%	48%	46%	46%	64%
Unable to work due to other reasons	7%	7%	5%	10%	9%

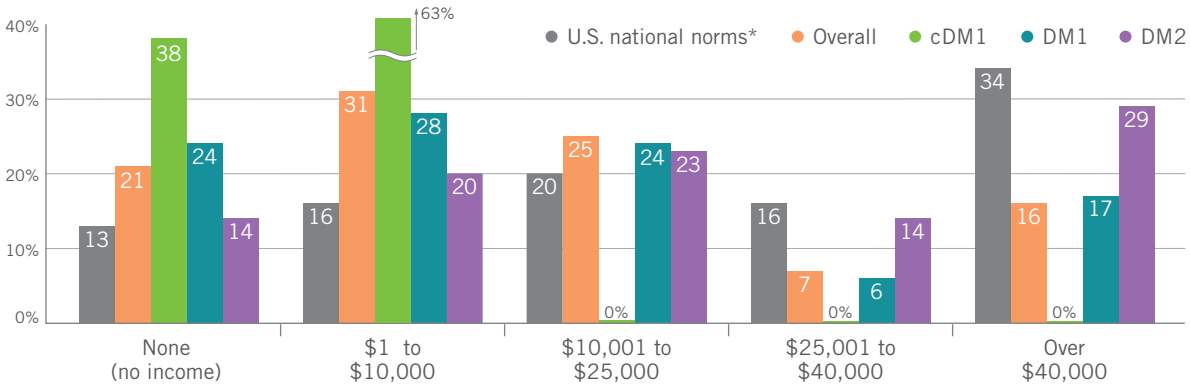
NOTE: Percentages can add up to more than 100% because multiple responses were permitted

Notably:

- Only 25% of **Overall** respondents aged 16 to 64 reported any level of employment at all: 14% fulltime; 11% part-time.
- 52% of **Overall** respondents aged 16 to 64 (including 64% of unknown/unsure (**U/U**) respondents) reported they were unable to work due to myotonic dystrophy.

Personal Income (U.S. respondents only)

Annual personal income findings are shown below for U.S. survey respondents aged 16 to 64, with comparison to the U.S. national norms. Findings for Canadian respondents were comparable but are not shown due to currency differences.



All numbers in percent (%), valid responses only

*Source: U.S. Census Bureau, 2015(b)

U.S. respondents reported considerably lower annual personal income compared to U.S. national norms.

Notably:

- 21% of **Overall** respondents reported they earned no annual income.
- 52% of **Overall** respondents earned less than $10,000 per year, compared to 29% nationally.
- 16% of **Overall** respondents earned more than $40,000 per year, compared to 34% nationally.
- 38% of **cDM1** respondents aged 16 to 64 earned no personal income at all.

Living Situation

	Overall	cDM1	DM1	DM2
I live alone	11%	0%	9%	16%
I live with my spouse/partner	55%	1%	63%	68%
I live with my child/children	18%	0%	26%	16%
I live with my parent(s)	26%	89%	22%	8%
I live with my sibling(s) and/or relative(s)	10%	37%	7%	4%
Other	6%	7%	3%	7%

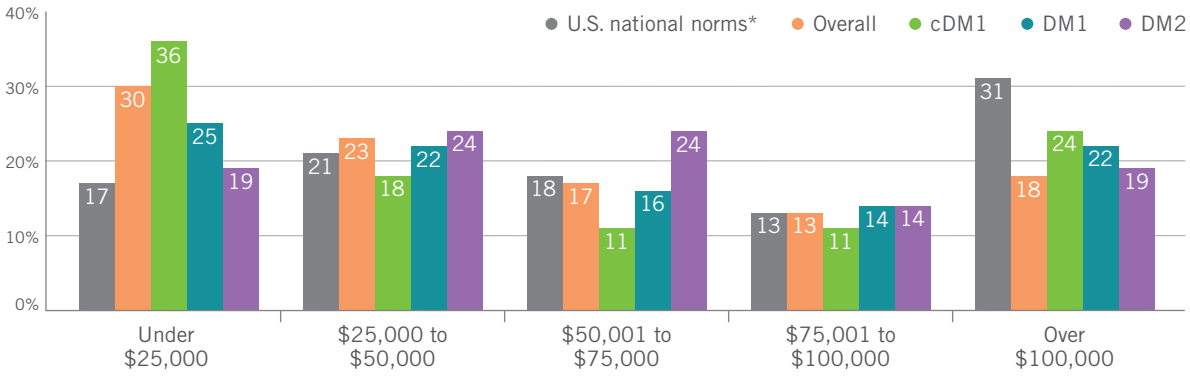
NOTE: Percentages can add up to more than 100% because multiple responses were permitted

Most respondents reported living in a traditional family unit. These results generally align with census data, except that only 11% of **Overall** respondents lived alone compared to 28% in the general population (U.S. Census Bureau, 2015(c); Statistics Canada, 2016(b)).

Significantly more female respondents reported they lived with their spouse/partner and/or children. Significantly more male respondents reported they lived with their parents (data by gender not shown).

Total Household Income (U.S. respondents only)

Total household income takes into account the combined income of all earners within a household. These findings are shown below for U.S. respondents, with comparison to U.S. national norms. Findings for Canadian respondents were comparable but are not shown due to currency differences.



All numbers in percent (%), valid responses only

*Source: U.S. Census Bureau, 2015(b)

U.S. respondents **Overall** and by type reported lower total household income compared to U.S. national norms:

Notably:

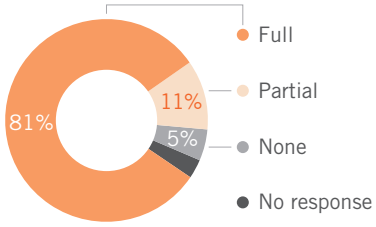
- 30% of **Overall** respondents reported total annual household income less than $25,000, compared to 17% nationally.
- 18% of **Overall** respondents reported total household income more than $100,000 per year, compared to 31% nationally.

Insurance Coverage

The United States and Canada have different systems of healthcare coverage (medical insurance). Results for each country are presented below:

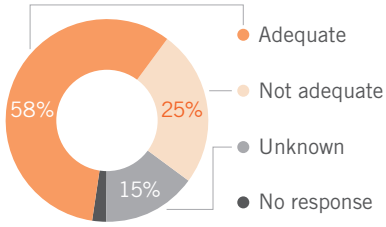
The United States Experience (n=944)

LEVEL OF COVERAGE



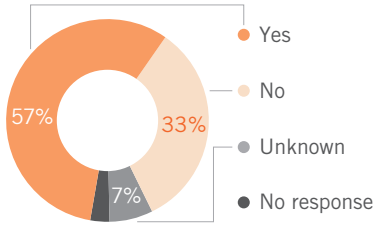
81% of U.S. respondents reported they had full health insurance coverage, 11% had partial coverage, and 5% had no coverage at all.

ADEQUACY OF COVERAGE



When asked if they felt their coverage was adequate, 25% reported it was not and 15% reported they didn't know.

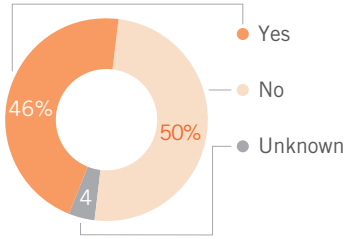
ABLE TO MEET COSTS



Further, 33% reported they experienced difficulty meeting the costs of their medical care.

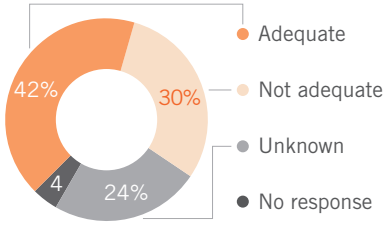
The Canadian Experience (n=236)

SUPPLEMENTAL COVERAGE



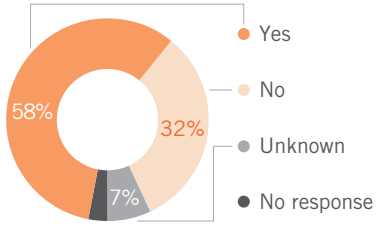
In Canada, all respondents have access to universal health care coverage. Almost half (46%) reported they also had additional (supplemental) insurance coverage.

ADEQUACY OF COVERAGE



When asked if they felt their coverage was adequate, 30% reported it was not and 24% reported they didn't know.

ABLE TO MEET COSTS



Further, 32% reported that they experienced difficulty meeting the costs of their medical care.

Biggest Obstacles in Managing Myotonic Dystrophy

When asked directly to identify the 'biggest obstacle' they faced in managing their myotonic dystrophy, most respondents (77%) described at least one specific obstacle. Their responses were grouped as follows:

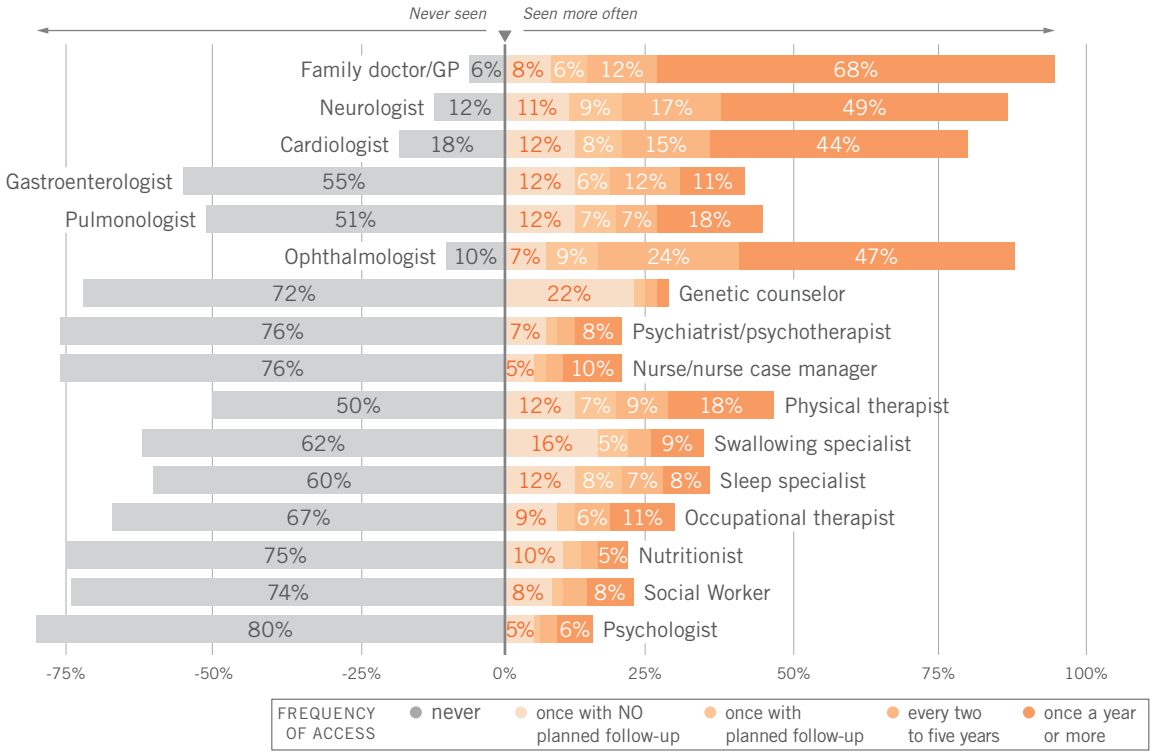
34%	Mobility-related obstacles like getting around, walking, balance, and accessibility issues
27%	Specific symptoms and physical problems, especially muscle weakness, pain, and gastrointestinal issues
23%	Household chores and daily tasks like cooking, cleaning, shopping, and running errands
21%	Sleep issues like daytime sleepiness, fatigue, and lack of energy
10%	Feelings and emotions like depression, anxiety, and guilt
8%	Cognitive impairments such as learning disabilities
4%	Lack of information and expertise about myotonic dystrophy
3%	Loss of independence including reliance on others and the need for support

Respondents were asked a series of questions about their healthcare-related experience in order to understand how they use the healthcare system, their satisfaction with it, and to identify unmet needs in access and awareness.

Healthcare Access

Each individual living with myotonic dystrophy has their own, unique healthcare needs. Specific providers should be seen accordingly.

FREQUENCY OF ACCESS TO HEALTHCARE PROVIDERS (OVERALL)



All numbers in percent (%), smaller numbers not shown

Frequency of Access

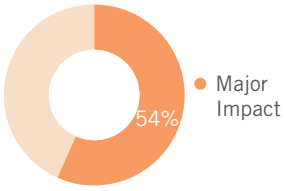
Primary healthcare providers (family doctors/GPs) were seen most frequently. Ophthalmologists, neurologists, and cardiologists were the most frequently seen secondary providers. Allied health professionals (such as occupational therapists and nutritionists) were seen considerably less often or not at all.

cDM1 respondents reported the most frequent and comprehensive visits to healthcare providers (data by type not shown):

- They saw primary and secondary providers significantly more often, particularly gastroenterologists, pulmonologists, and sleep specialists.
- They saw allied health professionals substantially more often, particularly social workers, physical therapists, nurse case managers, nutritionists, speech/swallowing specialists, and occupational therapists.

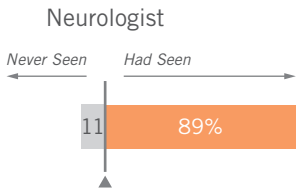
Assessing Needs in Healthcare Access

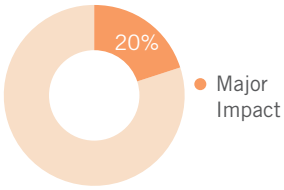
Survey results were analyzed to determine whether respondents who reported major impact from a particular group of symptoms were accessing relevant specialists.



Muscle Symptoms

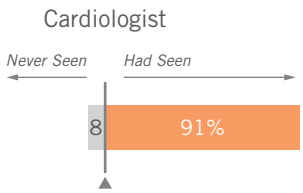
54% of overall respondents reported major impact from muscle symptoms of whom 11% had never seen a neurologist.

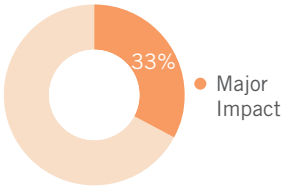




Cardiorespiratory Symptoms

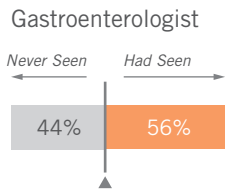
20% of overall respondents reported major impact from cardiorespiratory symptoms of whom 8% had never seen a cardiologist.*

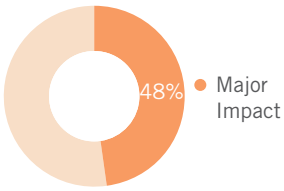




Gastrointestinal Symptoms

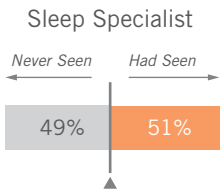
33% of overall respondents reported major impact from gastrointestinal symptoms of whom 44% had never seen a gastroenterologist.

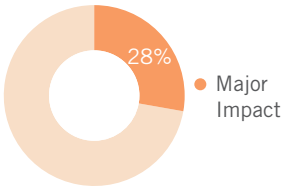




Sleep Symptoms

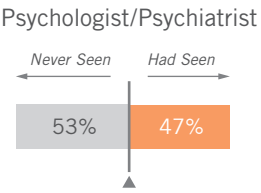
48% of overall respondents reported major impact from sleep symptoms of whom 49% had never seen a sleep specialist.





Psychological Symptoms

28% of overall respondents reported major impact from psychological symptoms of whom 53% had never seen a psychologist/psychiatrist.



*Cardiac screening is a necessary part of clinical care for anyone with myotonic dystrophy.

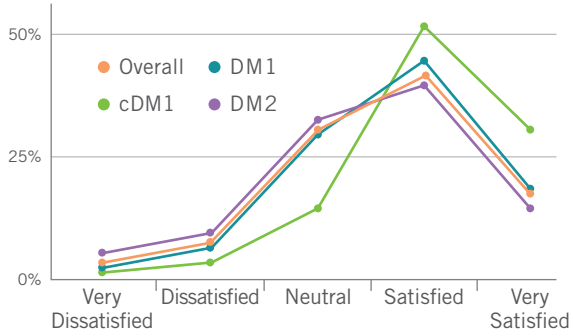
Most Helpful Healthcare Providers

Overall, respondents reported the most helpful providers were neurologists, followed closely by family doctors/GPs and cardiologists. **DM1** respondents identified cardiologists as the most helpful provider more often than other groups. The **cDM1** respondents identified physical therapists, occupational therapists, and speech/swallowing specialists as the most helpful providers more often than other groups.

Satisfaction with Healthcare

Only 10% of **Overall** respondents reported any level of dissatisfaction with the medical care they receive.

The **cDM1** group reported the highest level of satisfaction with medical care.



LEVEL OF SATISFACTION WITH HEALTHCARE

Unmet Medical Needs

Respondents were asked directly if they had specific medical needs or concerns that had not been met through their healthcare. Of those that offered a specific response (38%), the most frequently reported unmet needs or concerns involved:

Medical Expertise and Systems of Care
finding providers with knowledge and expertise in myotonic dystrophy, referrals to specialists, coordination of care, consistency of follow-up

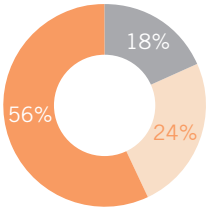
Information and Resources
good information and resources on the disease, its management and progression, including availability of treatments and interventions

Symptoms and Physical Problems
managing specific physical aspects of myotonic dystrophy, especially cardiac risks, pain, daytime sleepiness/fatigue, and gastrointestinal issues

Cardiac Risk Awareness

Cardiac (heart) problems can sometimes go undetected but have significant implications for quality of life and longevity in myotonic dystrophy patients. The survey asked about respondents' current awareness of the associated cardiac risks.

CARDIAC RISKS



18% indicated they were not aware of the cardiac risks associated with myotonic dystrophy.

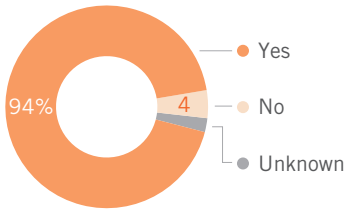
24% were aware but had not discussed these risks with a medical professional.

56% were aware of these risks and had discussed them with a medical professional.

Cardiac Screening

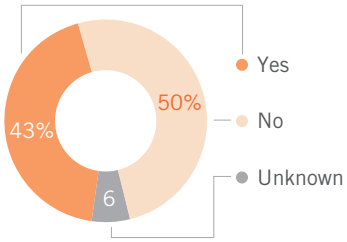
Managing risk through cardiac screening (heart testing) is a necessary part of clinical care.

CARDIAC TESTING PERFORMED



94% of **Overall** respondents reported they had their heart tested using an electrocardiogram (ECG or EKG), echocardiogram, or cardiac MRI.

ABNORMAL HEARTBEAT



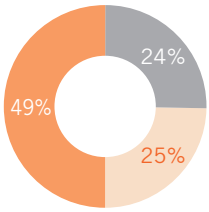
43% of **Overall** respondents reported they had been diagnosed with an abnormal heartbeat/ cardiac conduction problem.

It is critical that patients speak with a qualified healthcare professional about cardiac risks (conduction problems, abnormal heartbeat, and/or weakened heart muscle) and cardiac symptoms (chest pain, racing/fluttering heart, and/or dizziness/fainting) and receive appropriate medical care and advice accordingly.

Anesthesia Risk Awareness

Anesthesia poses a particularly high risk to myotonic dystrophy patients, with severe health implications. Survey respondents were asked about their current awareness of these risks.

ANESTHESIA RISKS



24% indicated they were not aware of the general anesthesia risks associated with myotonic dystrophy.

25% were aware but had not discussed these risks with a medical professional.

49% were aware of these risks and had discussed them with a medical professional.

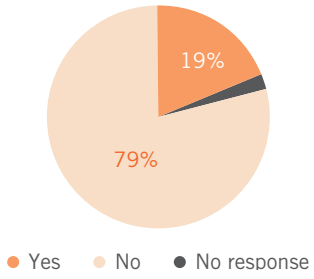
For a helpful guide on managing anesthesia risks visit: www.myotonic.org

Treatments and Medical Interventions

While there is no medication specifically approved for myotonic dystrophy, patients may be prescribed certain medications, devices, and/or interventions to help manage some specific symptom areas.

Medication Use by Major Symptom Area

If you have myotonia (difficulty relaxing muscles), do you take medication for it?



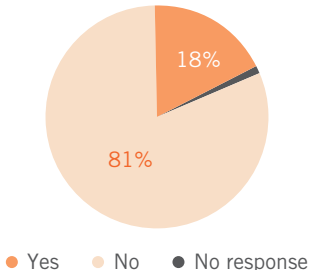
Of the respondents who had myotonia, 19% reported they were taking medication for it.

45 different medications were reportedly being used for myotonia. The most commonly reported was Mexiletine.

Satisfaction with medications being used for myotonia:
- 5% extremely satisfied
- 16% very satisfied
- 32% moderately satisfied
- 21% slightly satisfied
- 11% not at all satisfied
- (17% no response)

DM2 respondents were most likely to take medication for myotonia (24%).

If you have fatigue and/or daytime sleepiness (DTS), do you take medication for it?



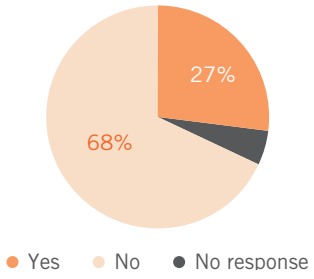
Of the respondents who had DTS and/or fatigue, 18% reported they were taking medication for it.

20 different medications were reportedly being used for DTS/fatigue. The most common were Modafinil and Methylphenidate.

Satisfaction with medications being used for DTS/fatigue:
- 7% extremely satisfied
- 28% very satisfied
- 28% moderately satisfied
- 10% slightly satisfied
- 7% not at all satisfied
- (20% no response)

DM1 respondents were most likely to take medication for DTS and/or fatigue (27%).

If you have gastrointestinal (GI) issues, do you take medication for them?



Of the respondents who had GI issues, 27% reported they were taking medication for them.

51 different medications were reportedly being used for GI issues. The most common were Polyethylene Glycol and Omeprazole.

Satisfaction with medications being used for GI issues:
- 6% extremely satisfied
- 30% very satisfied
- 39% moderately satisfied
- 12% slightly satisfied
- 2% not at all satisfied
- (10% no response)

cDM1 respondents were most likely to take medication for GI issues (34%).

The use of medications in myotonic dystrophy is complicated and some medications are potentially harmful. People with myotonic dystrophy should speak with a qualified physician to understand the potential risks and benefits associated with their use.

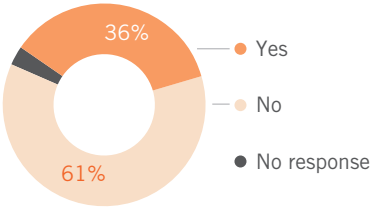
Specific Medical Interventions

Respondents were asked about their experiences with the following specific medical interventions:

Pacemakers and Defibrillators

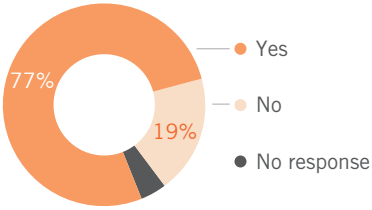
Sometimes doctors recommend a pacemaker and/or implantable cardioverter defibrillator (ICD) to help manage cardiac risks in myotonic dystrophy.

PACEMAKER/ICD RECOMMENDED



Of the 43% of **Overall** respondents who were diagnosed with an abnormal heartbeat (see *Cardiac Screening* section), 36% indicated their doctor recommended a pacemaker and/or ICD.

PACEMAKER/ICD USE

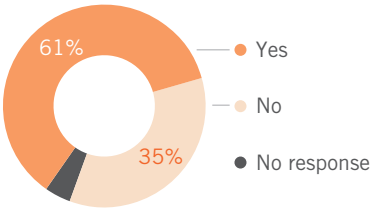


Of those who received the recommendation, 77% (140 people) went ahead with it. Reasons for not going ahead with it included timing, conflicting medical advice, and/or cost.

Breathing Devices

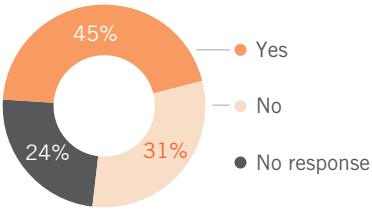
Sometimes doctors recommend an assistive breathing device (e.g., CPAP, BiPAP) to help manage sleep apnea (trouble breathing during sleep) and other similar symptoms.

CPAP/BiPAP RECOMMENDED



Of those respondents who experienced sleep apnea (see *Sleep and Fatigue Symptoms* above), 61% indicated their doctor recommended a CPAP or BiPAP machine.

CPAP/BiPAP USE



Of those who received the recommendation, 45% reported they currently used the device. The most common reason for not using it was inability to tolerate the mask while sleeping.

Of those who reported on their satisfaction with the use of a breathing device, 53% reported they were only ‘slightly’ or ‘not at all’ satisfied (data not shown).

Cataracts and Cataract Surgery

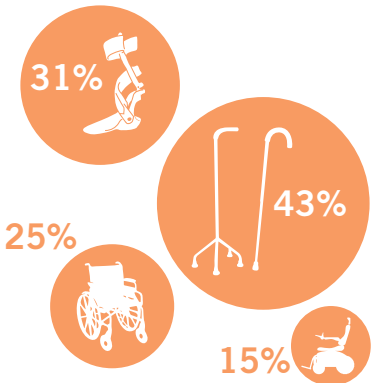
Cataracts are common in myotonic dystrophy patients and can significantly impair vision. There are simple surgical options available for their treatment.

EXPERIENCE WITH CATARACTS

	Overall	cDM1	DM1	DM2
Yes, I have had cataracts	54%	8%	56%	66%
And, yes, I have had cataract surgery	66%	50%	64%	69%
Average age of first cataract surgery (in years)	44	15	43	50

54% of **Overall** respondents have had cataracts, 66% of whom have had surgery to remove them. The survey did not explore why the others had not had cataract surgery.

Use of Mobility Devices



The most challenging activities for the majority of respondents were mobility-related (see *Managing Daily Life* section). Of the 92% of **Overall** respondents who reported mobility challenges, 58% indicated use of some sort of assistive device.

As illustrated, the most commonly used devices were canes, crutches, and walkers (43%), followed by leg and/or ankle braces (31%), manual wheelchairs (25%), and electric wheelchairs (15%).

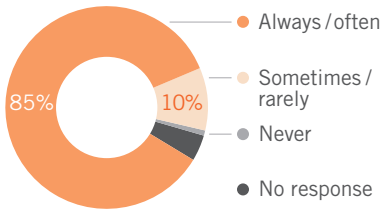
Reported satisfaction with mobility devices was relatively high, with only small numbers of respondents indicating that they were 'not at all helpful'.

Following Medical Advice

Recommended treatments and interventions can only be effective when patients follow the medical advice they receive (compliance).

85% of **Overall** respondents reported they 'always' or 'often' followed their doctors' advice (i.e., follow-up on referrals, fill and take prescriptions, interventions, etc.).

COMPLIANCE



Questions related to the use of medication, specific medical interventions, and the use of devices to manage myotonic dystrophy symptoms should be directed to a qualified healthcare professional.

Information and Resources

Access to information and opportunities for patient engagement can empower patients and families to take an active role in their own disease management.

Information Sources

Respondents were asked how they obtain information and what sources had been most helpful.

Computer Use and Information Access

64% of **Overall** respondents reported they used a computer/device to search the Internet and/or check email daily or weekly. When asked specifically how often they accessed information about myotonic dystrophy, 35% indicated they did so regularly (monthly or more).

Support Groups

Engaging in support groups, whether local or online, can provide mutual benefit to participants by allowing them to share their knowledge and learn from the experiences of others.

EXPERIENCE WITH SUPPORT GROUPS

	Overall	cDM1	DM1	DM2
I would 'not likely' attend a support group	45%	45%	39%	36%
Yes, I would attend 'if one was available'	48%	52%	49%	58%
I already attend my local support group	6%	1%	9%	5%
No response	2%	1%	2%	1%

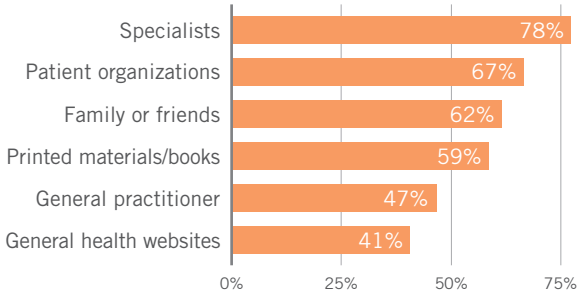
6% of **Overall** respondents indicated they currently attend a support group.

48% of respondents indicated they would attend a support group meeting 'if one was available' in their area.

Helpful Sources of Information

Respondents rated the helpfulness of specific sources of myotonic dystrophy information.

% WHO RATED SOURCE 'SOMEWHAT' OR 'VERY' HELPFUL



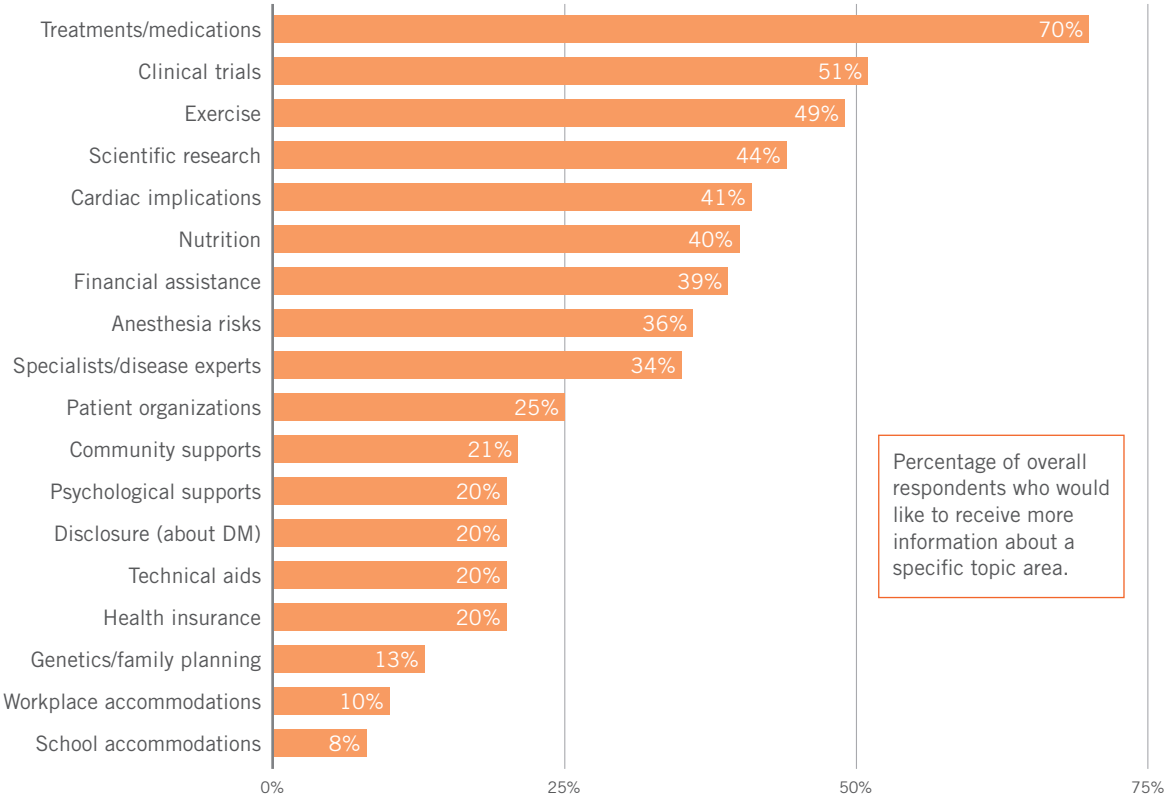
As shown, 78% of **Overall** respondents rated medical specialists as 'somewhat' or 'very' helpful information sources. Patient advocacy organizations were rated the second most helpful source, followed by family or friends.

The **cDM1** group rated medical specialists even higher, with 94% rating them as 'somewhat' or 'very' helpful. All information sources were reportedly less helpful for **DM2** respondents, with the exception of general health-related websites (data by type no shown).

Information Needs

Respondents were asked about their specific information needs including what they wanted to know more about and how they wanted to get the information.

WHAT RESPONDENTS WANTED TO KNOW MORE ABOUT



70% of respondents wanted to know more about available treatments/medications, 51% wanted to know more about clinical trials, and 49% wanted to know more about exercise. The **cDM1** respondents were much more interested than other groups in finding out more about school accommodations/assistance (data by type not shown).

PREFERRED WAYS TO RECEIVE INFORMATION (ranked)

	Overall	cDM1	DM1	DM2
Lettermail (printed)	61%	44%	60%	67%
Talking with health care providers	55%	62%	57%	47%
Reading books and other printed material	43%	44%	46%	42%
Email (electronically)	38%	41%	45%	49%
Visiting patient organization websites	36%	39%	46%	43%
Talking with friends/family	30%	25%	35%	23%
Attending support group meetings	18%	17%	20%	20%
Visiting other health-related websites	14%	10%	17%	19%
Participating in online forums/chat rooms	8%	13%	10%	15%

NOTE: Percentages can add up to more than 100% because multiple responses were permitted

Receiving information by lettermail (printed) was preferred almost two to one to receiving information by email (electronically). Further, speaking directly with a health care provider was a preferred method by over half of **Overall** respondents.

Patient Participation

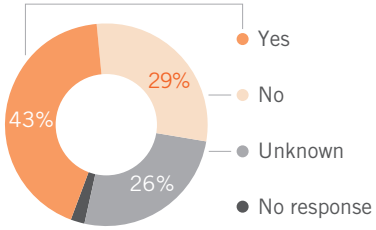
Patient participation in research is necessary to understand disease progression and its impact on health outcomes. Opportunities to become engaged include patient registries and research studies.

Registries

Patient registries serve as an important platform for organizing and informing patients as well as enabling research and clinical trials.

43% of **Overall** respondents indicated they were registered in a patient registry, 29% were not registered, and 26% reported they did not know if they were registered in a patient registry.

PATIENT REGISTRIES



WHAT IS A PATIENT REGISTRY?

A patient registry is a collection of standardized information about a group of patients who share a common condition or experience. The three largest myotonic dystrophy patient registries in the USA and Canada are:

- The National Registry for Myotonic Dystrophy (DM) and FSHD: urmc.rochester.edu/neurology/national-registry.aspx
- The Myotonic Dystrophy Family Registry: https://myotonicregistry.patientcrossroads.org
- The Canadian Neuromuscular Disease Registry: cndr.org

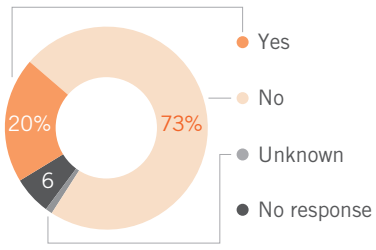
There are also a number of smaller registries at specific clinical locations in the United States and Canada and a number of international registries. Visit the links above and/or speak directly with a healthcare professional to learn more about these registries.

Research Studies

Research studies are another important investigative tool. They can include surveys, physician-led research studies (to learn about specific aspects of myotonic dystrophy), interventional studies (for testing a particular treatment or device), and clinical trials (for new drugs).

20% of **Overall** respondents reported they had been a part of a research study related to myotonic dystrophy.

RESEARCH STUDIES



– End of Section 3 –

Please refer to the *Key Findings* in Section 1 for summary results and highlights.
Additional information, including supplementary data tables, can be found at:
www.christopherproject.org

Section 4: Family Member/Caregiver Survey Results

In February 2015, 1,000 family member/caregiver surveys were mailed out to the original patient survey respondents who agreed to be contacted for further research. They were asked to give the survey to a family member or caregiver who helps them manage their myotonic dystrophy.

402 family members/caregivers responded to this follow-on survey. The feedback they provided is represented in **blue** across the following two themes:

About Family Member/Caregiver Respondents

- examines the role of family members/caregivers in supporting patients

The Patient Experience from the Family Member/Caregiver Perspective

- provides additional insight into the impact myotonic dystrophy has on patients and families

>>>

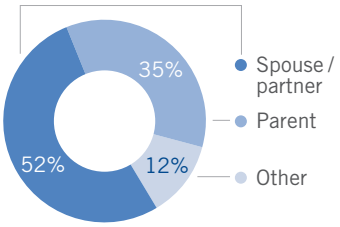
About Family Member/Caregiver Respondents

The first part of the family member/caregiver survey collected a profile of the family member/caregiver to understand who they are, their relationship to the patient, and the role they play in supporting the patient.

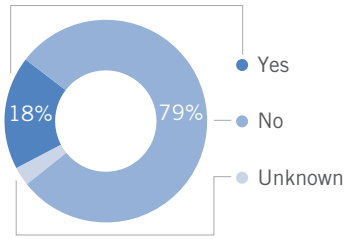
Family Member/Caregiver Profile

The large majority of family members/caregivers (85%) live with the person who filled out the original patient survey (95% for the **cDM1** group).

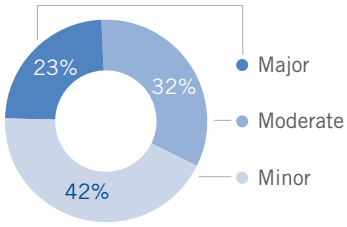
RELATIONSHIP TO PATIENT



HAS DM TOO?



LEVEL OF SUPPORT PROVIDED



As shown above, family member/caregiver respondents were mostly unaffected spouses or parents of the original patient survey respondent, who provided varying levels of support to the patient.

Family Member/Caregiver Demographics

- Average age of family members/caregivers: 57 years old (49 years for **cDM1** group)
- 51% were female, 47% were male
- 77% lived in the USA, 23% lived in Canada

Additional insights

Family member/caregivers were facing some major challenges:

- Nearly 1 in 5 had myotonic dystrophy themselves
- 23% reported they provided 'major' assistance to the patient
- 27% indicated they supported more than one person affected by myotonic dystrophy

Top 5 kinds of support provided by family members/caregivers

1. Emotional (87%)
2. Household tasks and chores (74%)
3. Attendance at clinical visits (68%)
4. Financial (56%)
5. Healthcare-related (55%)

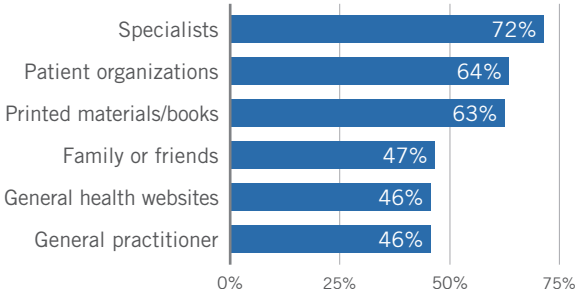
The majority of family members/caregivers provided patient support across multiple aspects of daily life and healthcare. Family members/caregivers of the **cDM1** group provided the most comprehensive support.

Family members/caregivers indicated emotional support, assistance with household tasks and chores, and helping with healthcare were the most helpful kinds of support they provided to the patient.

Information and Resources for Family Members/Caregivers

Family member/caregiver respondents rated the helpfulness of specific sources of myotonic dystrophy information.

% WHO RATED SOURCE 'SOMEWHAT' OR 'VERY' HELPFUL



As shown, 72% of family member/caregiver respondents rated medical specialists as 'somewhat' or 'very' helpful sources. Patient advocacy organizations were rated the second most helpful source, followed by printed materials and/or books. These ratings are consistent with the patient perspective.

Information Needs

Family members/caregivers were asked what they wanted to know more about and how they wanted to get the information:

Consistent with patient respondents, family members/caregivers were mostly interested in these top 5 topics:

1. Available treatments/medications (74%)
2. Clinical trials (55%)
3. Scientific Research (52%)
4. Exercise (49%)
5. Cardiac implications (46%)

Additionally, 38% of family members/caregivers wanted to know more about 'caregiver support/training'.

Family members/caregivers reported they wanted to receive information the same way patients did:

1. By lettermail (printed) (26%)
2. By email (electronically) (18%)
3. By talking with healthcare providers (10%)
4. Internet/online (10%)
5. By reading books and other printed material (8%)

Biggest Challenges for Family Members/Caregivers

Family members/caregivers were asked directly what their 'biggest challenge' was in helping the patient manage their myotonic dystrophy. 84% of family members/caregivers provided a detailed written response. Three major themes emerged:

Managing the Complexity of the Disease: 43%

43% reported that helping to manage a complex neuromuscular condition was their greatest challenge.

Examples:

- accessibility and mobility issues; getting around
- understanding and dealing with the broad spectrum of physical, emotional, and psychological challenges that come with myotonic dystrophy
- interacting effectively with medical professionals and the healthcare system

Family Member/Caregiver Burden: 33%

33% reported that the physical and emotional impact on the family member/caregiver themselves was their greatest challenge.

Examples:

- having the time, energy, and strength needed to provide care (especially when caregiver has myotonic dystrophy too) including the risk of exhaustion/burnout
- emotional impact of having myotonic dystrophy in the family like depression, frustration, and guilt; accepting reality; dealing with an uncertain future
- meeting costs of care; insurance costs; financial stress

Family members/caregivers of the **cDM1** group reported Caregiver Burden was their biggest challenge (44%).

Coaching & Supporting: 21%

21% reported that encouraging growth and enhancing quality of life for the patient was their greatest challenge.

Examples:

- encouraging positive behavior; providing motivation regarding lifestyle, diet, and exercise
- supporting patient in their own self-care; encouraging independence
- providing emotional support; helping patient deal with anxiety and fear
- maintaining empathy, understanding, and patience

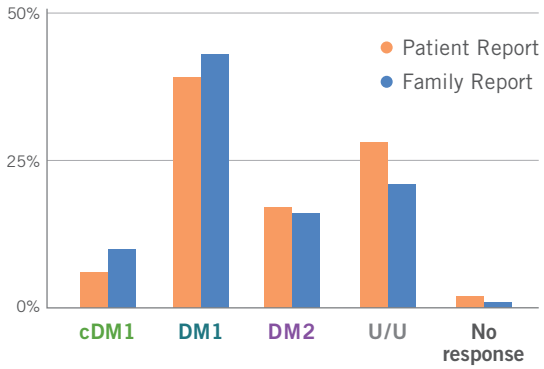
The Patient Experience from the Family Member/Caregiver Perspective

Family members/caregivers were asked some of the same questions that patients were asked across a variety of areas (e.g., the patient's type of myotonic dystrophy, the patient's general health, the patient's symptoms, and others). The responses they provided are compared (in **blue**) to the patients' responses (in **orange**) and are presented below.

The Patient's Type of Myotonic Dystrophy

As shown, each type of myotonic dystrophy experienced by patients is well-represented in the family members/caregiver respondent group.

There was general alignment between what patients reported and what their family members/caregivers reported. However, 32% did not report the same type of DM as each other: some reported a specific type when the other had selected unknown/unsure (**U/U**) and some reported a different type altogether.



PATIENT'S TYPE OF DM

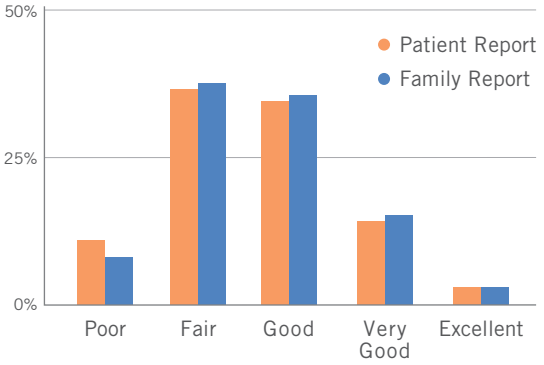
The Patient's Age of Symptom Onset and First Symptoms

	Overall Patient	Family Member/ Caregiver
Average age of onset	25.7 years	25.4 years
First symptoms noticed	Muscle weakness 38%	Muscle weakness 32%
	Myotonia 32%	Myotonia 21%

The patient's age of symptom onset and first symptoms reported by family members/caregivers were consistent with what patients reported about themselves.

The Patient's General Health

The results of the family member/caregiver report on the patient's general health were consistent with what the patient reported, as shown on the right.



PATIENT'S GENERAL HEALTH

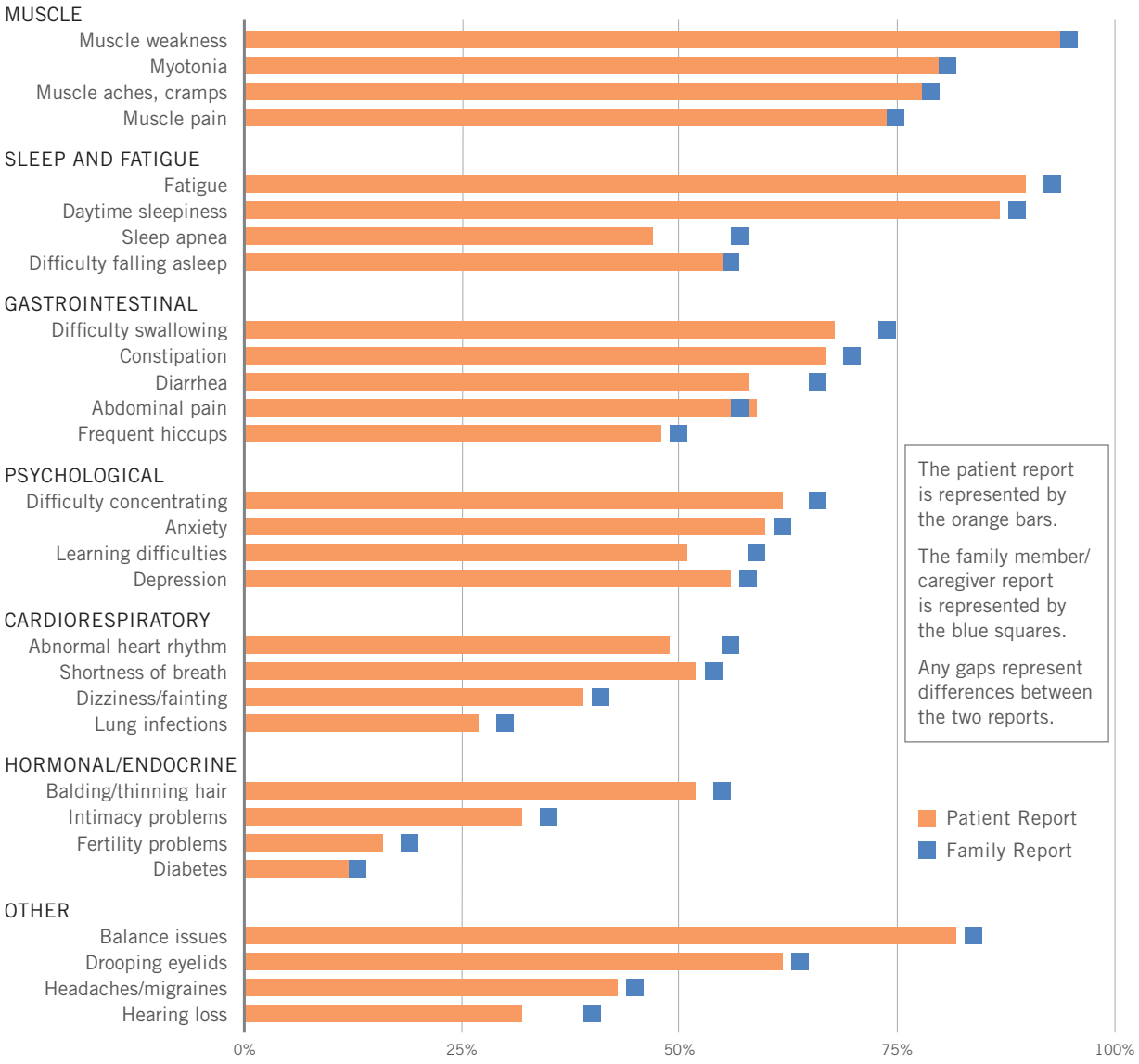
The Patient's Symptoms

To provide additional perspective on the patient report, family members/caregivers were also asked what symptoms the patient experienced and how much those symptoms impacted the patient's daily life.

The Patient's Symptom Prevalence

The percentage of family members/caregivers who reported the patient experienced a particular symptom (shown in **blue**) is compared to what the patients themselves reported (shown in **orange**).

PATIENT'S SYMPTOM PREVALENCE

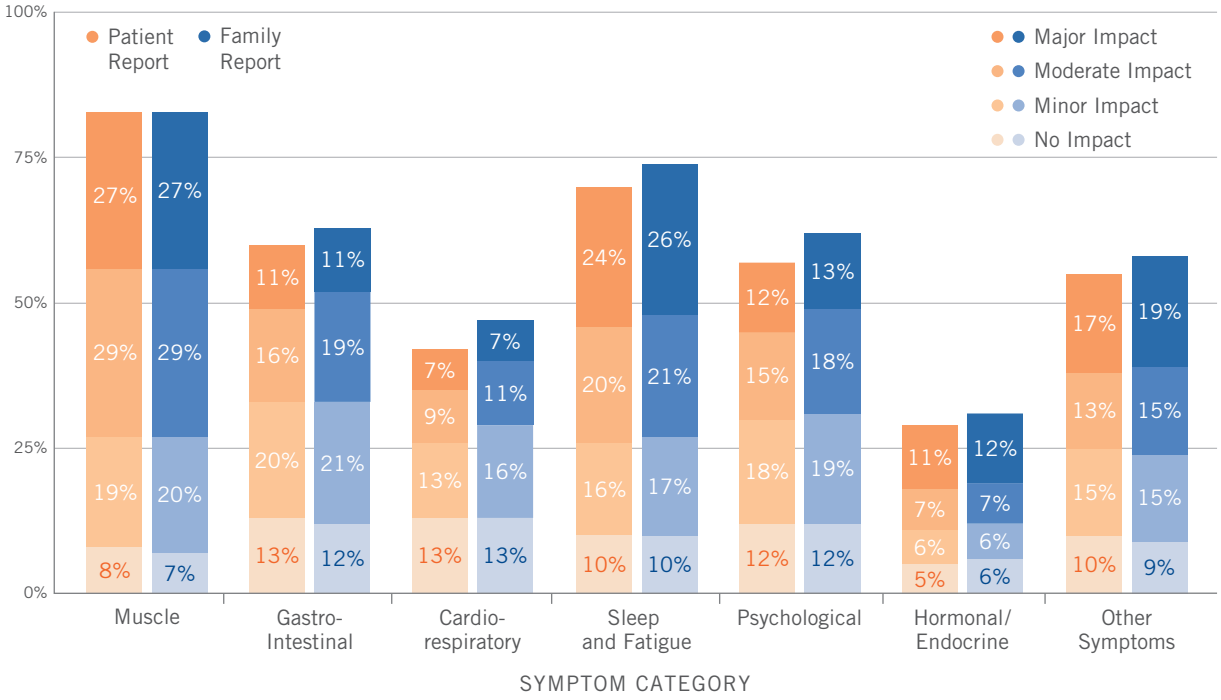


There is strong and consistent alignment in the symptom prevalence reported by patients and their family members/ caregivers. Generally, where there were any differences, family members/caregivers reported slightly higher symptom prevalence than patients themselves reported.

The Patient's Symptom Impact

Family members/caregivers also rated the level of impact each symptom or physical problem had on the patients' daily lives. Their ratings, grouped by category, are compared to what patients reported below:

PATIENT'S SYMPTOM IMPACT



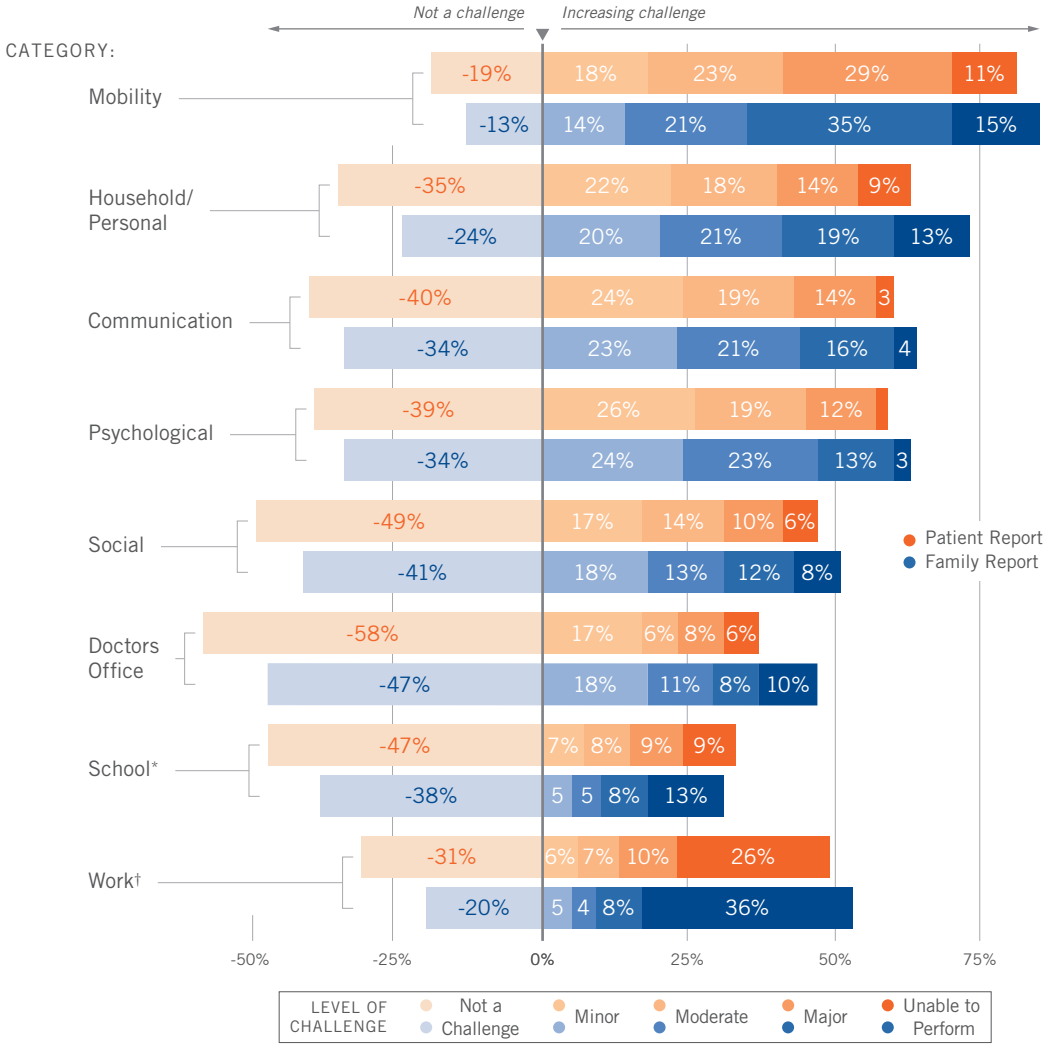
There is strong and consistent alignment in symptom impact reported by patients and their family members/caregivers. Generally, where there were any differences, family members/caregivers reported slightly higher symptom impact than patients themselves reported.

The Patient's Daily Life

Family members/caregivers were presented with the same list of 28 daily life activities as patients were asked to rate how much of a challenge they perceived each activity was for the patient to perform in daily life.

Their ratings were grouped by category and are presented in **blue** below with the patient report in **orange** for comparison:

PATIENT'S DAILY LIFE ACTIVITY CHALLENGES



All numbers in percent (%), smaller numbers not shown

*Only 80% of patients and 70% of family members/caregivers provided a rating for this item
†Only 79% of patients and 74% of family members/caregivers provided a rating for this item

Just like patients, family members/caregivers reported that people living with myotonic dystrophy face significant challenges in performing activities throughout their daily life.

While there was general agreement in which activities were the most challenging for the patient to perform, family members/caregivers consistently reported that patients experienced somewhat greater challenges than patients themselves reported.

The Patient's Biggest Obstacles

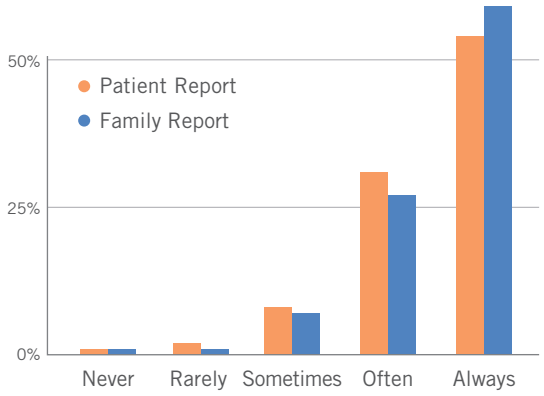
When family members/caregivers were asked directly to describe the '*biggest obstacle*' patients face in managing myotonic dystrophy, 79% reported at least one specific obstacle that significantly impacted the patient's daily life.

Reported by Patient		Family Member/ Caregiver
34%	Mobility-related obstacles like getting around, walking, balance, and accessibility issues	28%
27%	Specific symptoms and physical problems, especially muscle weakness, pain, and gastrointestinal issues	26%
23%	Household chores and daily tasks like cooking, cleaning, shopping, and running errands	6%
21%	Sleep issues like daytime sleepiness, fatigue, and lack of energy	18%
10%	Feelings and emotions like depression, anxiety, and guilt	17%
8%	Cognitive impairments such as learning disabilities	16%
4%	Lack of information and expertise about myotonic dystrophy	2%
3%	Loss of independence including reliance on others and the need for support	3%

Both groups reported mobility as the greatest obstacle and managing symptoms and physical problems as the second greatest obstacle in managing myotonic dystrophy in the patient's daily life. Far fewer family members/caregivers indicated that household chores/daily tasks presented a major obstacle for patients than patients did themselves. More family members/caregivers reported that psychological issues (both emotional and cognitive) presented a big obstacle for patients.

The Patient's Compliance with Medical Advice

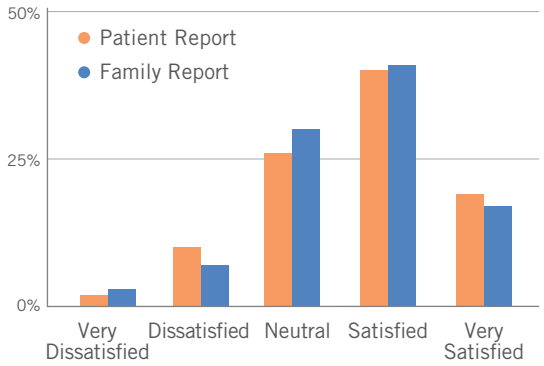
86% of family members/caregivers reported patients 'always' or 'often' follow their doctors' advice (i.e., referrals, prescriptions, interventions, etc.). This is consistent with the 85% of patient respondents who reported the same.



PATIENT'S COMPLIANCE WITH MEDICAL ADVICE

Satisfaction with the Patient's Medical Care

59% of family members/caregivers and 58% of patients reported that they are 'satisfied' to 'very satisfied' with the medical care the patient received.



SATISFACTION WITH PATIENT'S MEDICAL CARE

The Patient's Use of Medication for Daytime Sleepiness and/or Fatigue

17% of family members/caregivers reported that patients used medication for daytime sleepiness (DTS) and/or fatigue, which is consistent with the patient report. They also reported similar levels of satisfaction with the effect of these medications.

The Patient's Unmet Medical Needs

Family member/caregiver respondents were asked directly what specific medical needs or concerns they feel the patient had that had not been met through the patient's healthcare. Of those that offered a specific response (47%), the most frequently reported unmet needs or concerns involved:

Medical Expertise and Systems of Care
finding providers with knowledge and expertise in myotonic dystrophy, referrals to specialists, coordination of care, consistency of follow-up

Information and Resources
good information and resources on the disease, its management and progression, including availability of treatments and interventions

Symptoms and Physical Problems
managing specific physical aspects of myotonic dystrophy, especially cardiac risks, pain, daytime sleepiness/fatigue, and gastrointestinal issues

These unmet needs and concerns are consistent with what patients identified, except family members/caregivers cited Symptoms and Physical Problems more often than patients.

– End of Section 4 –

Please refer to the *Key Findings* in Section 1 for summary results and highlights.
Additional information, including supplementary data tables, can be found at:
www.christopherproject.org

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Christopher Project Reference Group

Past and current members:

Diane Bade: Patient Advocate
Marie-Hélène Bolduc: Muscular Dystrophy Canada
Elizabeth Florence: Myotonic Dystrophy Foundation
Paul Formaker: Myotonic Dystrophy Foundation
Cynthia Gagnon: GRIMN/University of Sherbrooke
Katharine A. Hagerman: Stanford University
Chad R. Heatwole: University of Rochester Medical Center
Leah Hellerstein: Myotonic Dystrophy Foundation
Sharon Hesterlee: Myotonic Dystrophy Foundation
Sarah Howe: Marigold Foundation
Don MacKenzie: Marigold Foundation
Sue McIntyre: Hindsight Group
Lianna Orlando: Muscular Dystrophy Association
John Porter: Myotonic Dystrophy Foundation
Marla Spiegel: Muscular Dystrophy Canada
Kristin Stephenson: Muscular Dystrophy Association
Eric T. Wang: University of Florida
Jodi Wolff: Muscular Dystrophy Association

Data Management and Analysis

All data gathered through the Christopher Project were managed under an IRB-approved research study protocol in accordance with the United States Federal Policy for the Protection of Human Subjects ('Common Rule') and the Canadian Tri-Council Policy Statement for Research Involving Humans (TCPS2) using best practices for privacy and data security.

Data Management: All surveys received were de-identified and checked for data quality. Qualitative data was grouped and coded according to identifiable themes. All data were then entered into a secure database using SPSS statistical analysis software. Range checking and summary statistics were used to ensure the quality of the aggregated dataset.

Data Analysis using SPSS: Descriptive statistical analyses were performed (frequency counts and measure of central tendency like averages) to summarize the results of all survey questions. Appropriate analytical techniques and statistical tests (i.e., Pearson's chi-square, column proportion tests, column means tests, t-tests, and ANOVA) were performed to test for significant differences between survey groups (i.e., types of myotonic dystrophy) and significant relationships between variables (i.e., age of onset vs. age of diagnosis, use of healthcare providers by symptom impact). Differences are only noted if they were observed to be statistically significant at a 99% confidence interval with a p-value of <0.01.

Data Rounding: Findings throughout this report are presented as whole percentages and, as such, are subject to small rounding errors.

Data Access

The extensive body of data gathered through the Christopher Project establishes a valuable and lasting resource for the entire myotonic dystrophy community and represents opportunities for qualified researchers and interest groups:

- to perform additional analysis of the existing dataset,
- to complement and compare it with other datasets, and
- to use it as a basis to support and inform further research.

Additional information, including supplementary data tables, can be found at:
www.christopherproject.org

Inquiries regarding access to the anonymized Christopher Project dataset can be made to:
support@christopherproject.org

Study Limitations

As with any population-based study, The Christopher Project is subject to certain limitations as described below:

- **Sampling bias:** The sample was recruited from patient advocacy group mailings lists, which represent only a subset of all the people living with myotonic dystrophy in Canada and the USA, and only a limited response rate (30-40%) was achieved.
- **Scope:** The depth of questioning in certain topics areas was limited in order to permit a broader examination of the patient experience.
- **Subjective report:** The study relied upon self-reporting from patients and family members/caregivers, some of which required them to recall past experiences and events.

Myotonic Dystrophy Resources

(selected, USA and Canada)

National Patient Advocacy Organizations:

- Muscular Dystrophy Association (USA): www.mda.org
- Muscular Dystrophy Association of Canada: www.muscle.ca
- Myotonic Dystrophy Foundation (MDF): www.myotonic.org

Patient Registries:

- The National Registry for Myotonic Dystrophy (DM) and FSHD: www.urmc.rochester.edu/neurology/national-registry.aspx
- The Myotonic Dystrophy Family Registry: https://myotonicregistry.patientcrossroads.org
- The Canadian Neuromuscular Disease Registry: www.cndr.org

Care Guidelines:

- Anesthesia Guidelines: www.myotonic.org (SEARCH 'Anesthesia')
- Consensus-based Care Recommendations for Adults with DM1: www.myotonic.org (SEARCH 'Care Recommendations')

Genetic References:

- OMIM Myotonic Dystrophy Type 1: www.omim.org/entry/160900
- OMIM Myotonic Dystrophy Type 2: www.omim.org/entry/602668
- Genetic Home Reference Myotonic Dystrophy: www.ghr.nlm.nih.gov/condition/myotonic-dystrophy
- GeneReviews® Myotonic Dystrophy Type 1: www.ncbi.nlm.nih.gov/books/NBK1165
- GeneReviews® Myotonic Dystrophy Type 2: www.ncbi.nlm.nih.gov/books/NBK1466

Other:

- Myotonic Dystrophy Toolkit: www.myotonic.org (SEARCH 'Toolkit')
- Stanford Neuromuscular Biobank: www.med.stanford.edu/day-lab/biobank.html
- Clinical Trials Database: www.clinicaltrials.gov (SEARCH 'Myotonic Dystrophy')

Additional copies of this report can be found online at: www.christopherproject.org

For more information or additional copies:

www.christopherproject.org

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