OneVOICE:

Insights and observations from a national survey
of adults and families living with neuromuscular disease

MDA® Muscular Dystrophy Association

INTRODUCTION: Why this study and why now?

This is an unprecedented moment for neuromuscular disease. The Food and Drug Administration (FDA) has approved eight new treatments in the last four years—some of them the first of their kind—and promising therapies are moving through the drug development pipeline. With this development we are seeing transformational opportunity to identify and treat individuals earlier—some as early as birth. For example, two neuromuscular disorders (Pompe and spinal muscular atrophy) have now been added to the national Recommended Uniform Screening Panel, a list of diseases for which the Secretary of Health and Human Services recommends every baby born in the U.S. be screened at birth. Newborn screening allows for early diagnosis, which leads to earlier treatment—and that means better health outcomes.

MDA wants to accelerate progress and remove barriers.

As the umbrella organization for more than 40 neuromuscular diseases, the Muscular Dystrophy Association (MDA) is committed to transforming the lives of people living with neuromuscular disease through supporting research to find the breakthroughs that will lead to treatments and cures, as well as providing programs to enrich patients' lives. Listening to the needs of the community is critical to MDA's mission to accelerate progress and remove barriers for patients and families. To that end, we sponsored a nationwide survey of patients and families to hear firsthand about their experiences and their unmet needs. How do they manage their disease day-to-day? What is their treatment burden? What trade-offs do patients and families make? Where do they get the information they need? These are the questions that will help us ensure that the programs and supports that are most needed are understood.

The study revealed a number of critical needs and opportunities. It also confirmed just how essential the programs and services provided by MDA are to the community, from MDA's network of multidisciplinary Care Centers, providing best-in-class care at more than 150 of the nation's top medical institutions, to our Resource Center, which serves the community with one-on-one specialized support, and our clinical trials finder that helps connect patients to trials.

This study is only the beginning of a continued commitment to listening to the community and doing our utmost to transform the lives of neuromuscular patients and their families.

Lynn Vos
President and CEO

STUDY OVERVIEW

MDA commissioned Edge Research, an established research firm with extensive expertise working with patient advocacy organizations, to conduct an objective and thorough study among the neuromuscular disease community. Specifically, the survey was composed of more than 50 questions that sought to identify the needs of individuals and families living with neuromuscular disease. The survey uncovered opportunities to achieve improved disease management and care as well as barriers for the neuromuscular disease community. Topics included in the survey comprised:

- Day-to-day needs and challenges of patients and caregivers
- Experiences around disease management
- Treatment benefits and risks
- Sources of support/service providers
- Information sources and needs/gaps today
- Interest in informational and support resources and services

This survey was distributed through MDA's national network of individuals living with or caring for someone with neuromuscular disease, as well as through local affiliates and partner organizations. In 30-days, more than 3,000 individuals provided complete responses to the survey, including 2,430 adult patients and 932 caregivers/family members (of which 727 were parents). More on the survey methodology is included at the end of this report.

Figure 1. Breakdown of respondents by type of neuromuscular disease

Type of Neuromuscular Disease (select all that apply)	Total respondents	% of total respondents
ALS (amyotrophic lateral sclerosis)	225	7%
Andersen-Tawil syndrome	5	0.1%
Becker muscular dystrophy (BMD)	165	5%
Central core disease	26	1%
Centronuclear myopathy	16	0.5%
Charcot-Marie-Tooth disease (CMT)	451	13%
Congenital muscular dystrophies (CMD)	128	4%
Congenital myasthenic syndromes (CMS)	17	1%
Dermatomyositis	29	1%
Duchenne muscular dystrophy (DMD)	365	11%
Emery-Dreifuss muscular dystrophy (EDMD)	27	1%
Facioscapulohumeral muscular dystrophy (FSHD)	251	7%
Friedreich's ataxia (FA)	30	1%
Giant axonal neuropathy (GAN)	3	0.1%
GNE myopathy/Nonaka myopathy/Hereditary Inclusion-Body myopathy (HIBM)	11	0.3%
Hyperkalemic periodic paralysis	4	0.1%
Hypokalemic periodic paralysis	13	0.4%
Inclusion body myositis	137	4%
Laing distal myopathy	7	0.2%
Lambert-Eaton myasthenic syndrome (LEMS)	9	0.3%
Limb-girdle muscular dystrophies (LGMD)	363	11%
Markesberg-Griggs late-onset distal myopathy	3	0.1%
McArdle disease	8	0.2%
Mitochondrial myopathy	73	2%
Miyoshi myopathy	10	0.3%
Myasthenia gravis (MG)	158	5%
Myofibrillar myopathy	13	0.4%
Myotonia congenita	20	1%
Myotonic dystrophy (DM)	289	9%
Myotubular myopathy	8	0.2%
Nemaline myopathy	11	0.3%
Oculopharyngeal muscular dystrophy (OPMD)	34	1%
Paramyotonia congenita	11	0.3%
Polymyositis	38	1%
Pompe disease	25	1%
Potassium-aggravated myotonia	1	0.03%
Scapuloperoneal myopathy	5	0.1%
Spinal muscular atrophy (SMA)	263	8%
Spinal-bulbar muscular atrophy (SBMA)	12	0.4%
Udd myopathy/tibial muscular dystrophy	1	0.03%
Other	196	6%
Diagnosis is not yet confirmed	59	2%
Not sure	24	1%

KEY FINDINGS

1. **Access to affordable, specialized care is a critical need.** The cost of care emerges as a *major* challenge for half (51%) of those who took the survey, and another third (31%) list it as minor concern. This has a real impact on the medical care those coping with neuromuscular disease receive. It's extremely troubling when 1 in 3 (33%) report that they or their loved one has delayed seeking or receiving medical attention because of the cost. In addition, access to *specialized care* is a pain point: 76% have concerns about access to health care and/or a health care provider with experience in neuromuscular disease, while also showing strong interest in securing these specialized services.

2. **Many patients still do not have a genetic diagnosis to confirm which disease they are living with.** Nearly a third (29%) of adult patients surveyed report that they have not had their diagnosis confirmed through genetic testing. Yet interest in doing so is strong. Age is a key factor, with the likelihood of being tested steadily decreasing with age. So too is cost, with nearly half (49%) of the community agreeing that genetic testing is expensive. And the community needs greater education around genetic testing: Open-ended responses reveal that the purpose and advantages of genetic testing today are not well understood by some patients/families, and even health care providers.

3. **The community wants more information and access to clinical trials.** Three quarters (76%) of the neuromuscular disease community have never participated in a clinical trial, yet interest is high. Most of those who have not participated (63%) would consider doing so, and clinical trials top the list of topics where the community wants more information. The biggest barriers to participation are not being asked about or not knowing about clinical trials. Lower-income, less educated patients and caregivers are interested in trials but less likely to report enrollment in clinical trials and significantly less likely to say that they feel informed about clinical trials happening.

4. **Mobility/independence is top of mind for the neuromuscular disease community.** Independence and mobility are major concerns for most (64%) of the community, topping the list of more than 60 different types of issues or situations that people living with neuromuscular disease and their families face. Adult patients tend to have a positive outlook regarding their own independence and productivity, translating into high interest in services and resources that aid in this, particularly disability-modified vehicles, accessible housing, and travel solutions. Caregivers are more divided in their outlook, with half concerned about their loved one's ability to live an independent life. The emotional and physical toll of caregiving comes through in the survey's open-ended feedback.

5. **MDA is a go-to resource.** The community is eager for information and support and counts on MDA as their number one resource on neuromuscular disease. Topping the list of services from MDA are: the MDA Resource Center, specialized medical care, help finding clinical trials, and assistance with independent living. The survey shows that these, along with ways to connect with others living with these diseases, are even more important to the most vulnerable, under-served patient populations. Yet notable gaps exist between interest and awareness, highlighting an important need to amplify awareness of these in-demand resources.

DETAILED FINDINGS

Those living with neuromuscular disease face a variety of challenges every day, from small inconveniences to major barriers that impact physical, social, and emotional wellbeing. To better identify and prioritize concerns of the community, the survey asked participants to rate their level of concern on more than **60 different issues** or situations that people living with neuromuscular disease and their families might face. These fell into five different categories:

- Physical health
- Financial and insurance
- Lifestyle/employment/education
- Emotional/relationships
- Access/adherence

As illustrated in Figure 2 below, physical health and financial concerns rise to the top across audiences, specifically concerns around independence, mobility, the cost of medical devices and equipment, as well as uncertainty about the future. Physical health issues – the ability to walk and have the energy to make it through the day – weigh most heavily on patients. The cost of and access to home modifications are more top of mind for caregivers. These concerns reveal several key areas of need within the community which MDA and its partners are poised to meet.

Figure 2. Top 10 major concerns by audience

Top Concerns: Mobility, Cost of Devices, Uncertainty about Future

Top 10 Patient Concerns*

1. **64% Independence regarding mobility/getting around**
2. 62% Ability to walk (ambulation)
3. **55% Uncertainty about the future**
4. 53% Having the energy to make it through the day
5. **53% Cost of medical devices and equipment**
6. **50% Access to health care/ provider experienced in ND**
7. **49% Cost of medical care**
8. **47% Activities of daily living (bathing,dressing, etc.)**
9. 45% Loss of income
10. 45% Cost of prescriptions and treatments

Top 10 Caregiver Concerns*

1. **68% Uncertainty about the future**
2. **63% Independence regarding mobility/getting around**
3. **62% Cost of medical devices and equipment**
4. 58% Avoiding and managing contractures
5. 57% Cost of home modifications
6. **56% Activities of daily living (bathing, dressing, etc.)**
7. 56% Housing issues (home modifications/accessibility)
8. **56% Access to health care/ provider experienced in ND**
9. 54% Respiratory complications
10. **53% Cost of medical care**

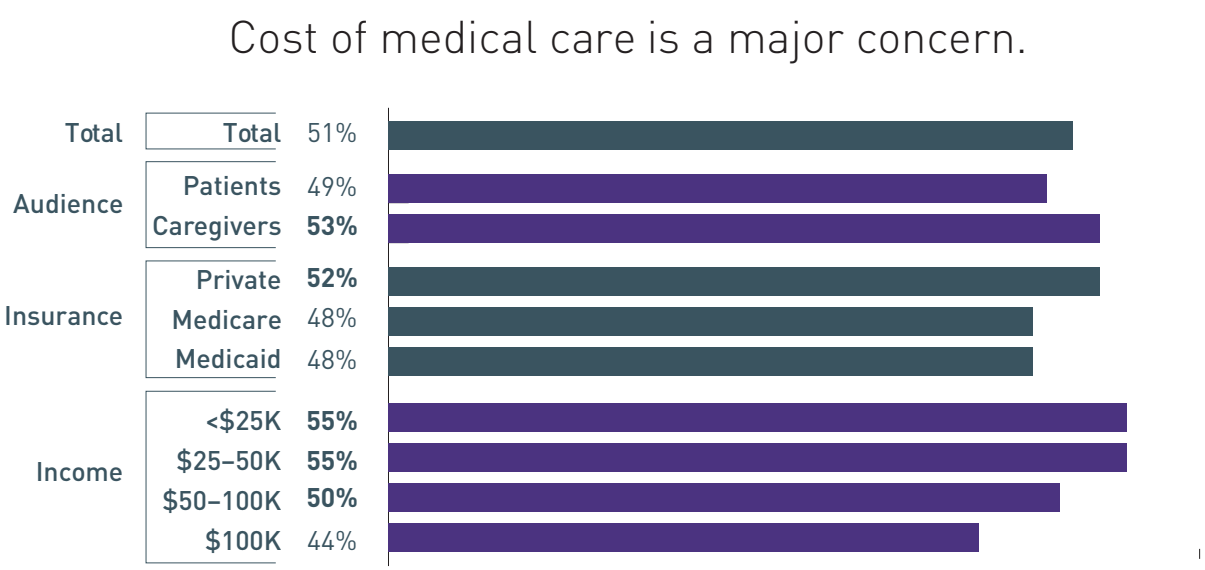
Bolded items on both lists

* "Major concerns" out of 64 issues total

Finding 1: Access to affordable, specialized care is a pain point and critical need.

The cost of medical care is a well-documented challenge for many patients and families living with neuromuscular disease.[1] Indeed, more than half of the community reports that the cost of medical care is of *major* concern (51%), another third (31%) say it's a minor concern. Cost concerns tend to be higher among low-income patients (income under $50k), those who live in the South, and those who are paying for private insurance (vs. Medicare/Medicaid recipients). Moreover, roughly 1 in 3 (33%) report they have delayed seeking or receiving medical attention in the past year due to concerns around cost. This is particularly true with CMT patients (46%).

Figure 3: Concern about cost of medical care. (Bolding indicates statistical significance.)

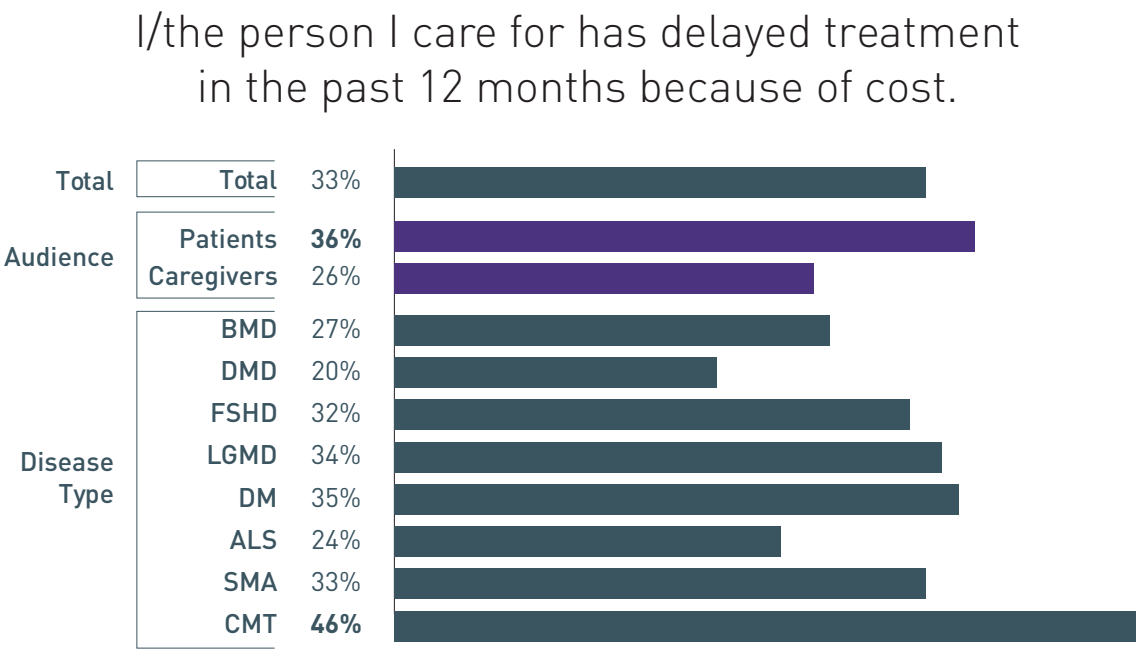


1. Larkindale J et al. Cost of illness for neuromuscular diseases in the United States. Muscle Nerve. 2014 Mar;49(3):431-8. https://www.ncbi.nlm.nih.gov/pubmed/23836444

   Thayer S, Bell C and McDonald CM. *The Direct Cost of Managing a Rare Disease: Assessing Medical and Pharmacy Costs Associated with Duchenne Muscular Dystrophy in the United States.* J Manag Care Spec Pharm. 2017 Jun;23(6):633-641. https://www.ncbi.nlm.nih.gov/pubmed/28530521

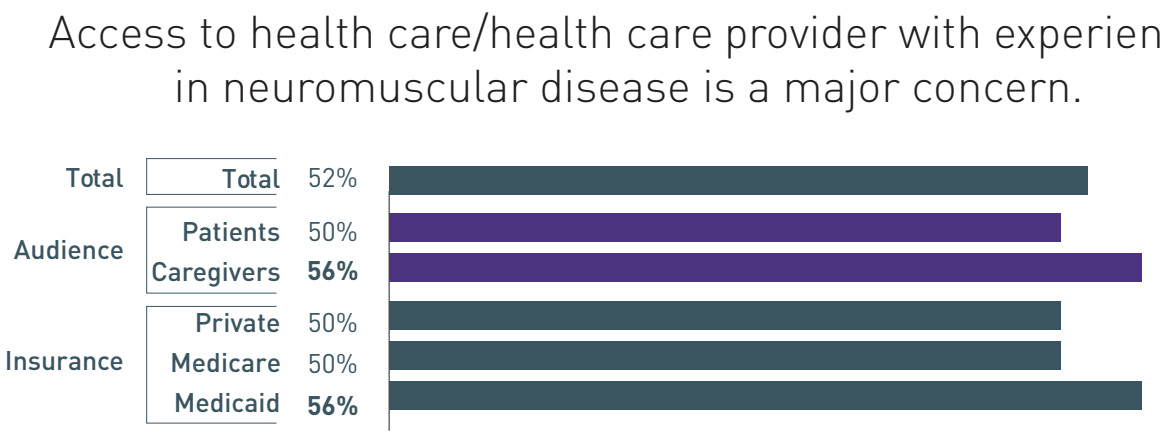
   Capkun G et al. *Burden of illness and healthcare resource use in United States patients with sporadic inclusion body myositis.* Muscle Nerve. 2017 Nov;56(5):861-*867.* https://www.ncbi.nlm.nih.gov/pubmed/28493327

Figure 4: Medical care delayed because of cost. (Bolding indicates statistical significance.)



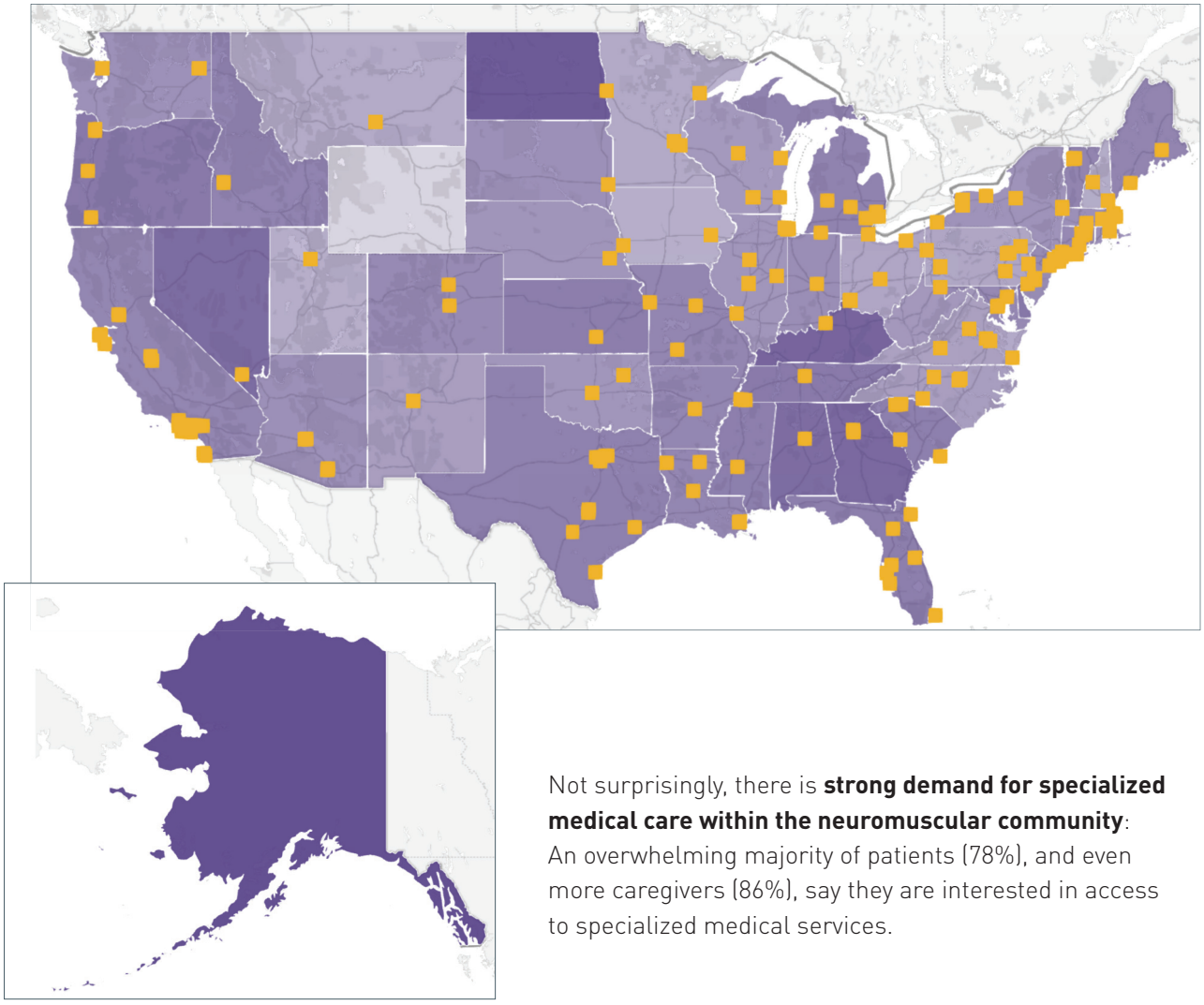
Beyond cost, access to care and specialized medical care is a major challenge. Around half (52%) of respondents have major concerns about access to health care and/or a health care provider with *experience in neuromuscular disease*, and another 24% say this is a minor concern. Access to specialized care is particularly concerning for Medicaid recipients (56% major concern) and adult patients ages 30-49 (56%). This concern applies across disease types, although ALS patients show slightly lower levels of concern (just 39% say it is a "major concern").

Figure 5: Concern about specialized care. (Bolding indicates statistical significance.)



Access to specialized care is inconsistent across the United States. Patients and caregivers in specific states, including Alaska, Nevada, Kentucky, Alabama, and Georgia show the most concern about access to health care providers with experience in neuromuscular disease (more than 65% cite this as a major concern). These states also have just 3 or fewer MDA Care Centers each, all located in urban centers. Eighty percent of Alaskans expressed this concern and there is no MDA Care Center in their state. Conversely, a state like Pennsylvania has 12 MDA Care Centers and concern is much lower.

Figure 6. Concern about specialized care varies across the country (overlaid with care center locations)



Darker purple=greater concern

Yellow=MDA Care Center location

Not surprisingly, there is **strong demand for specialized medical care within the neuromuscular community**: An overwhelming majority of patients (78%), and even more caregivers (86%), say they are interested in access to specialized medical services.

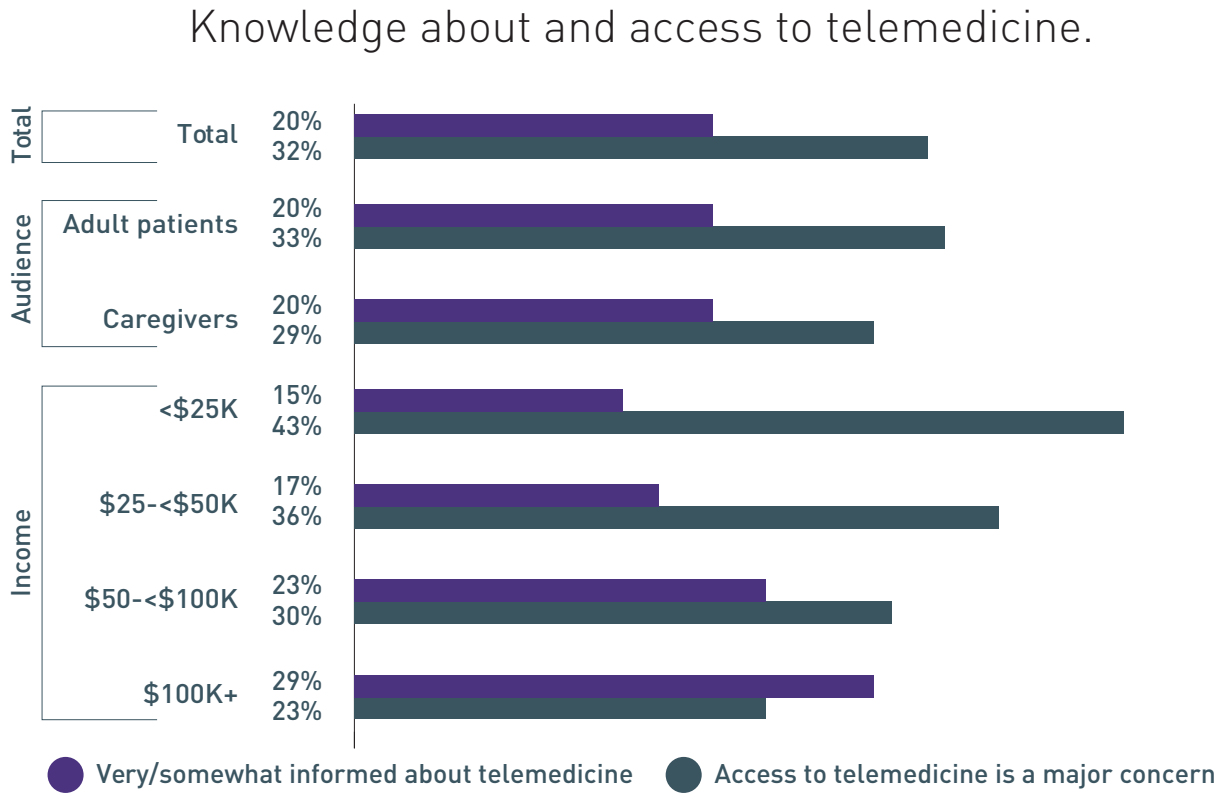
Solutions: Specialized Medical Care

No matter where one lives across the U.S., there are resources to receive specialized care for neuromuscular disease:

- **MDA Care Centers:** MDA's Nationwide Care Center Network is the largest network of neuromuscular clinics, providing best-in-class care at 150+ of the nation's top medical institutions. The Care Center Network conducts 90,000+ medical visits annually for more than 60,000 individuals living with muscular dystrophy, ALS, and related neuromuscular diseases, and includes more than 2,000 clinical providers. MDA Care Centers also serve as a hub of neuromuscular research activity with 12,000+ individuals participating in clinical trials and natural history studies across the network in 2017 alone. MDA's Care Centers and Affiliates offer an important opportunity for many in the community to access specialized services and are currently utilized by about half of the community who responded to this survey: 53% say they have visited a center. The Care Center Network offers tens of thousands of appointments each year for individuals living with muscular dystrophy, ALS, and other neuromuscular diseases so they may access expert multidisciplinary care, clinical trials, and connect with MDA and the neuromuscular community.

- **Telemedicine:** For those in more rural or remote areas of the country, or without easy access to a Care Center, telemedicine offers a viable solution for cost-effective, real-time interaction and communication with specialists. However, this service is **not widely utilized nor understood by the community today**. Only 20% say they feel informed about telemedicine, while 1 in 3 (32%) say access to telemedicine is a major concern. Lower-income patients have higher levels of concern about access and report being less informed about telemedicine.

Figure 7: Few are informed about telemedicine.



Finding 2. Many patients still don't have a genetic diagnosis.

Today, genetic testing is available for many patients living with neuromuscular disease.

Genetic testing is not only used to confirm a diagnosis but also can help predict disease course, inform recurrence risk, and facilitate enrollment into clinical trials. However, a quarter (25%) of survey respondents say they or their loved one has not had their diagnosis confirmed through genetic testing, and this rises to a third (29%) among adult patients.

Interest in testing, however, is high across all populations. When those who have not been tested were presented with a list of possible reasons, only 4% said this was because they did not want testing. What is deterring patients from getting tested?

Age is a factor. As shown on the following page in Figure 9, likelihood of receiving a genetic test decreases dramatically with age, with almost 4 in 10 (38%) adult patients over age 50 reporting that they have not received a genetic test. Open-ended responses suggest this may be due to the availability of this technology at the time of their diagnosis: e.g. *"I was diagnosed with CMT before genetic testing was available"* – Adult patient. Indeed, older patients (including 54% of those over 65) are significantly more likely to say they did not receive testing because it was "not offered" compared to younger patients.

Figure 8: A quarter have not received a diagnosis confirmation through genetic testing.

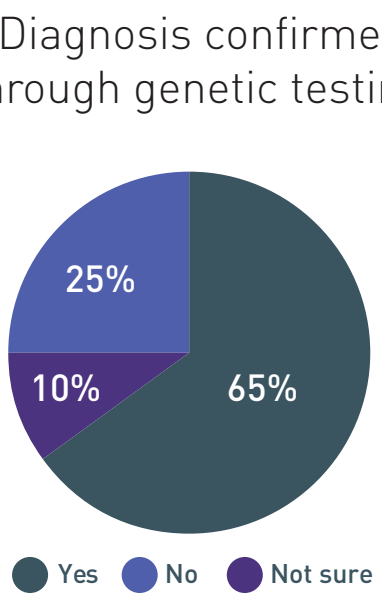
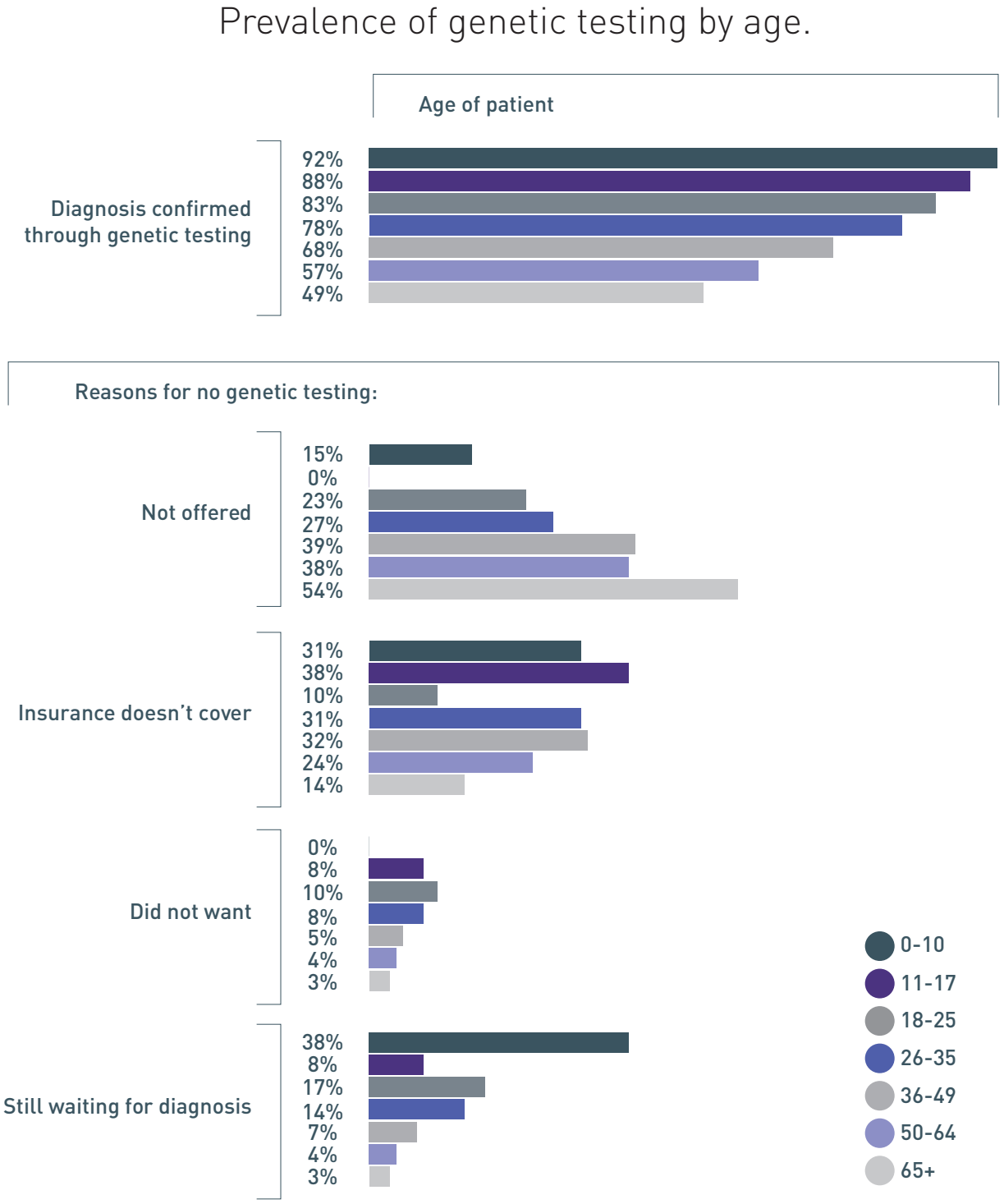


Figure 9: Likelihood of genetic testing decreases dramatically by age



Disease type is also a factor. ALS, FSHD, and CMT patients are significantly less likely to have received testing compared to other disease types, which may be due in part to differences in the availability of tests across diseases.

Another barrier is cost. Half of participants (49%) feel genetic testing is not affordable, and a quarter (22%) of those who did not get a genetic diagnosis cite "lack of insurance coverage."

Many respondents list "other" reasons for not receiving a genetic diagnosis. Analysis of open-ended data uncovers three common barriers: (1) a genetic test was conducted but results were negative or inconclusive; (2) diagnosis was confirmed using a different type of test (e.g. blood test, muscle biopsy, symptoms); and/or (3) a genetic test was considered unnecessary due to a family history of the disease. All together, these responses suggest that the purpose and advantages of genetic testing are not always understood by the community.

Indeed, nearly 1 in 3 (29%) say they do not feel informed about genetic testing. Adult patients (particularly those over 70), patients diagnosed more than 10 years ago, and low-income patients/caregivers with less than a college education are significantly less likely to feel informed.

Figure 10: Genetic diagnosis varies by disease type

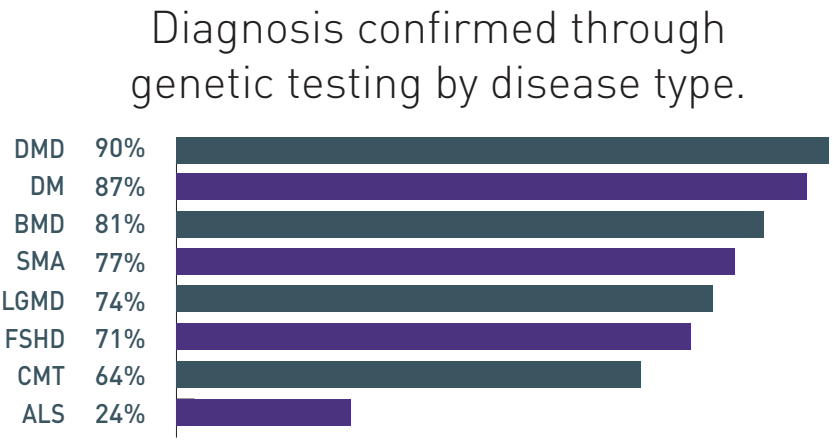
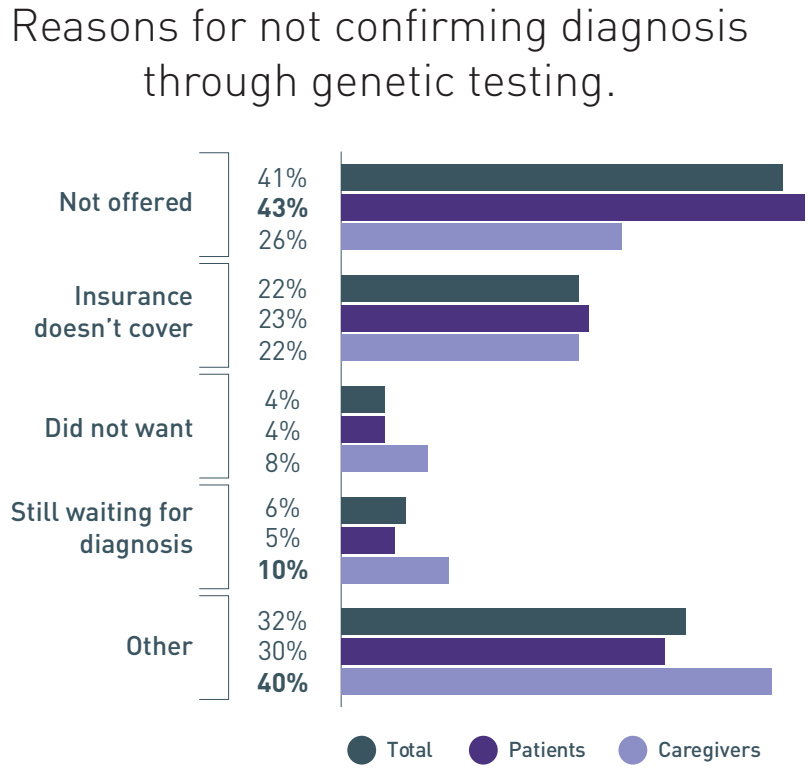


Figure 11: Barriers to genetic testing. (Bolding indicates statistical significance.)



Open-ended feedback reveals that patients and physicians may be misinformed around the costs, coverage, and advantages of genetic testing

Survey Question:

What are some reasons why you or the person you care for has not had a confirmation through genetic testing?

"If there's no cure, no treatment, and I was not having children - why do it?"
– Adult patient

"My brother and mother were both confirmed to have FSHD through genetic testing, so I felt it wasn't necessary for me to have it as well. I was diagnosed by my neurologist based on my symptoms and their tests."
-Adult patient

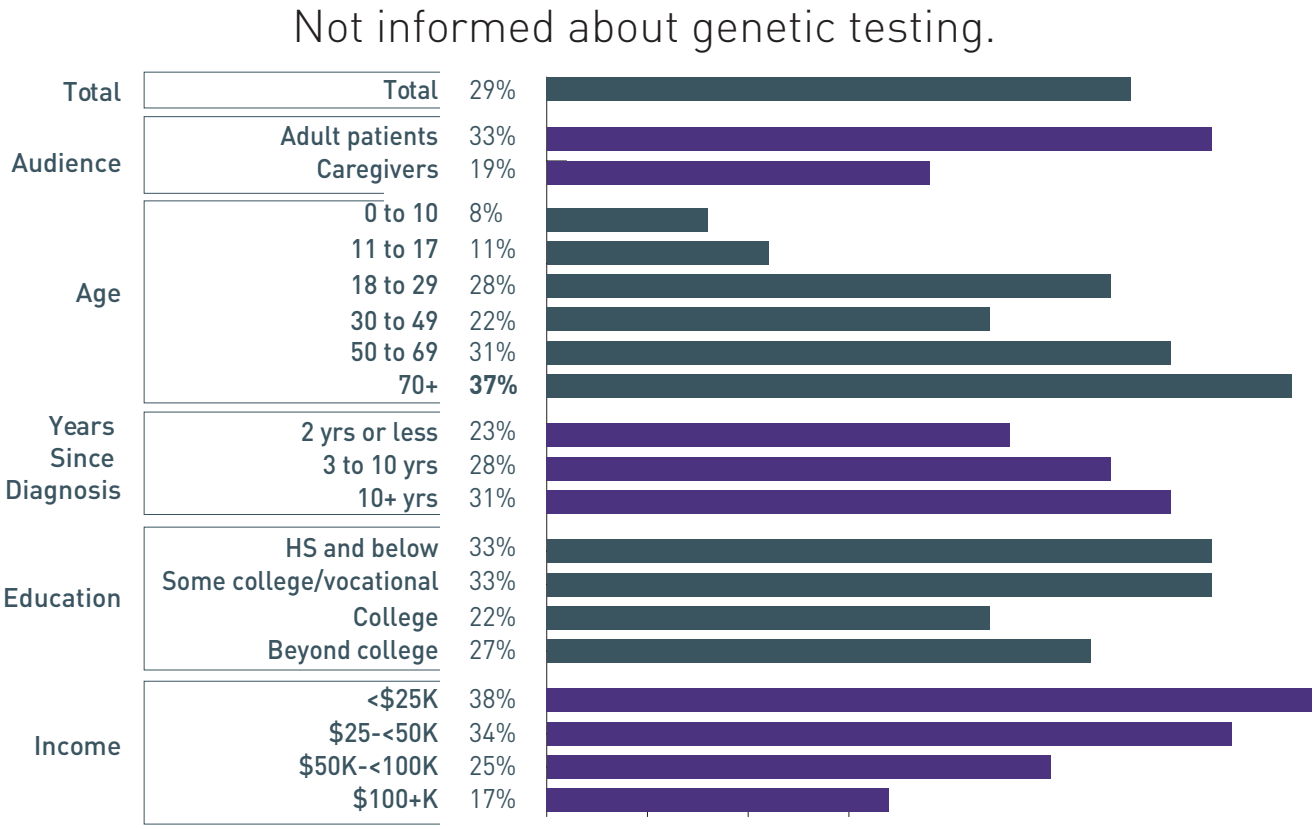
"Myasthenia gravis is not genetic."
-Adult patient

"The doctors didn't think it was genetic. No family history."
- Caregiver

"The doctor stated my insurance will not cover genetic testing due to my mitochondrial myopathy diagnosis."
– Adult patient

"The doctor said it cost $10,000 which I don't have."
– Adult patient

Figure 12: Several audiences are less informed about genetic testing. (Bolding indicates statistical significance.)



The Benefits of Genetic Testing

Genetic testing is available to many neuromuscular patients. Beyond confirming a diagnosis, testing results are used to target therapies and facilitate enrollment into clinical trials. However, many neuromuscular patients, caregivers, and health care providers may not have all the facts when it comes to the cost and benefits of genetic testing. Patient advocacy organizations can play an important role in educating the community around genetic testing and why it is important. In particular, the neuromuscular disease community needs to know:

- Genetic testing is widely available and has been vastly improved. Those with previously inconclusive or inaccurate test results can get a more accurate diagnosis today.
- Genetic testing uncovers the molecular basis for the disease, which can lead to more targeted therapy. Getting tested or re-tested can improve accuracy of diagnosis by pinpointing an individual's disease-causing mutation, which in turn can lead to eligibility for certain therapies or medications and clinical trials as well as a better understanding of how the disease will progress.
- Genetic testing is becoming more affordable.

MDA's MOVR (neuroMuscular ObserVational Research) Data Hub gathers clinical and genetic data combined with patient-reported data from patients who agree to share their information with their physician at an MDA Care Center. This large dataset provides researchers and clinicians with insights into the relationship between genes (genetic makeup) and disease characteristics and outcomes to enhance disease understanding, optimize health outcomes, and aid the development of more targeted drugs and therapies. All data is saved in a secure database and de-identified — meaning all personal information is protected. State-of-the-art security measures protect patient privacy, yet also provide for dynamic aggregation of data for research purposes and the ability for researchers to reach out to clinicians about trials and advances that may benefit individual patients of theirs.

Finding 3: The community wants more information and access to clinical trials.

Clinical trials play a key role in the discovery and development of new treatments for neuromuscular diseases. However, three quarters (76%) of the neuromuscular disease community have never participated in a clinical trial. Younger patients (ages 11-17) and DMD and ALS patients are most likely to be currently or previously enrolled in a clinical trial, and patients diagnosed within the last 10 years are more likely to currently be enrolled in a trial than those with an earlier diagnosis.

Among those who have not participated, interest in clinical trials is high: 63% would consider doing so, while just 13% are not interested. Interest in trials is highest among adult patients under 70 and transcends disease types, with the exception of DMD and ALS, which have a higher availability of clinical trials compared to other diseases. And, clinical trials top the list of 23 different topics related to neuromuscular disease that patients and caregivers are most interested in learning more about.

Figure 13: Clinical trial participation.

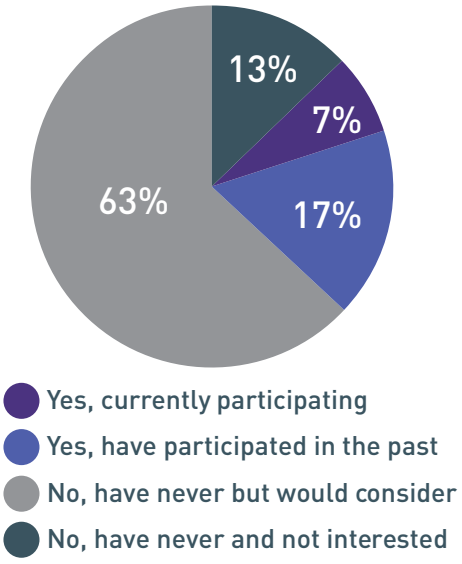
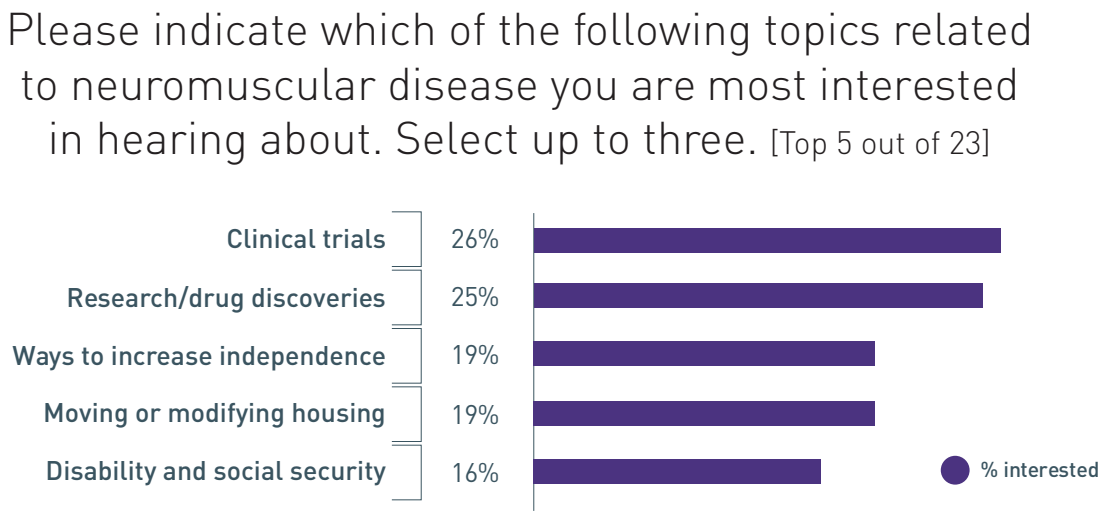
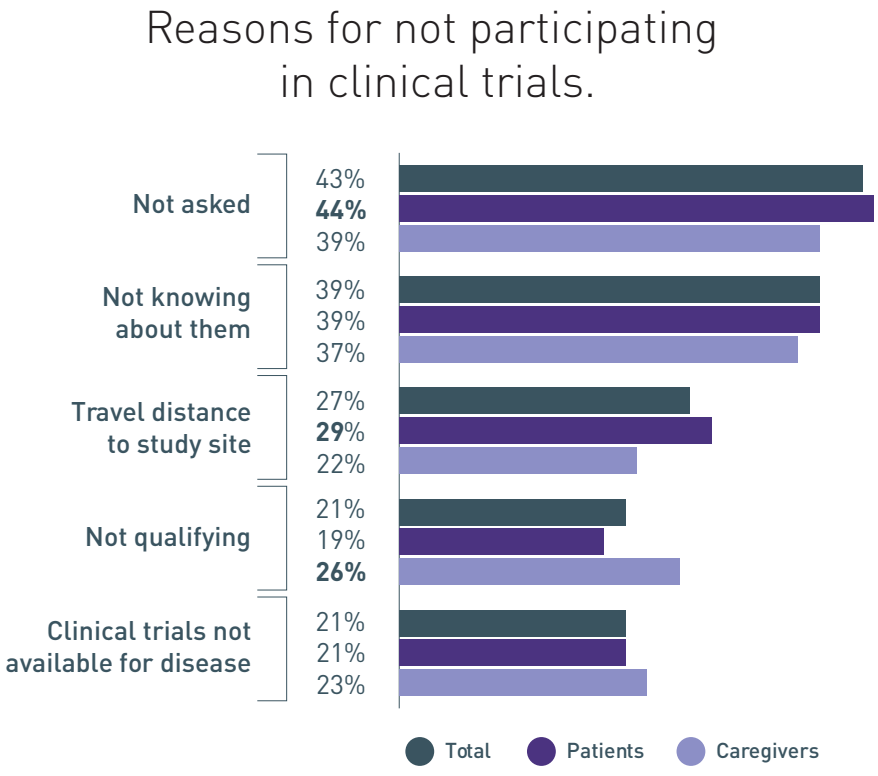


Figure 14. Top 5 topics of interest related to neuromuscular disease.



What is responsible for this discrepancy between interest and participation? Barriers to participation echo those reported for genetic testing: "not being asked" tops the list (43% among those who have not participated), followed by "not knowing about them" (39%). These sentiments cut across most demographics – income, education, age, and race/ethnicity – however, adult patients diagnosed more than 10 years ago and those with a diagnosis of LGMD and DM are slightly more likely to cite these barriers.

Figure 15: Barriers to clinical trial participation. (Bolding indicates statistical significance.)



Some survey respondents took time to write in additional barriers to participation. Analysis of these responses provides insight into why patients aren't being asked or have low awareness: namely, that many patients rely heavily on their doctors to initiate communication and follow up, as well as provide guidance around eligibility and cost for participation. When these conversations don't happen, or doctors themselves are misinformed, then patients are reluctant to follow up themselves.

Doctors are key to clinical trial participation

Survey question: ***What are some reasons why you/the person you care for has not participated in a clinical trial?***

"We were told he could participate but never heard anything else from the doctor to sign him up."
-Caregiver

"There was one in our area I was interested in, but the physician responsible for evaluating participants never initiated it."
– Adult patient

"The doctor didn't follow through with the request, then he left clinic. No continuity of care."
– Caregiver

"I've had a doctor tell me that I couldn't afford to have the stem cells done."
– Adult patient

"My doctor does not think the data is strong enough."
– Adult patient

Roughly 1 in 5 (21%) report that they had not participated because they did not qualify for a trial. A review of the open-ended responses highlights that lack of genetic testing is a problem for some, e.g. *"I applied but did not qualify because I haven't had a diagnosis via genetic testing"* – Adult patient. For others, this may be due in part to concerns that they are too old: *"Assumption that I am too old, and the treatments/trials are only for people that who have hope of preventing further disability."*- Adult patient

Other research suggests that low-income and minority populations are generally more skeptical of clinical trials.[2] Indeed, patients and caregivers in this survey with higher incomes and education levels are more likely to be/have been enrolled in a clinical trial. But this seems to be about access and information, not interest. Barriers cited by low-income and minority populations did not dramatically differ from other populations with the exception that lower-income patients and caregivers are more likely to cite travel distance (31% say a barrier) and financial concerns (22%) compared to higher-income groups. In addition, low-income, less educated caregivers and patients are also significantly less likely to report they feel informed about clinical trials. While most are interested in assistance with finding clinical trials, interest levels are slightly lower compared to those of higher-income, more educated patients and caregivers.

2 Durant RW et al. Perspectives on Barriers and Facilitators to Minority Recruitment for Clinical Trials among Cancer Center Leaders, Investigators, Research Staff and Referring Clinicians. Cancer. 2014 April 1; 120(0 7): 1097–1105.

Durant RW et al. Different Types of Distrust in Clinical Research Among Whites and African Americans. J Natl Med Assoc. 2011 February ; 103(2): 123–130.

Taylor AL and Wright J. Importance of Race/Ethnicity in Clinical Trials. Circulation. 2005;112:3654-3666.

Figure 16: Socio-economic status impacts access and information about clinical trials, but not interest.

Interest and participation in clinical trials.

	Total	Education				Income			
		HS or less	Some college	College	Post-graduate	<$25K	$25-50K	$50-100K	$100K+
Currently participating	7%	4%	6%	9%	9%	3%	6%	8%	**12%**
Participated in the past	17%	13%	16%	20%	20%	14%	14%	19%	22%
Never participated, but interested	63%	63%	66%	61%	61%	67%	65%	63%	60%
Never participated and not interested	13%	18%	13%	10%	11%	14%	15%	10%	7%
Reasons for not participating:									
Travel distance to study site	27%	22%	31%	26%	24%	31%	33%	25%	20%
Financial concerns	15%	14%	18%	17%	9%	22%	18%	15%	5%
% not informed	41%	45%	45%	37%	38%	50%	44%	40%	30%
% interested in help finding clinical trials	79%	76%	77%	82%	82%	79%	77%	81%	85%

Solutions: Raising Awareness of Clinical Trials

The community is clamoring for more information about clinical trials. The majority in this survey say they want help finding clinical trials: **79% say they are interested in "assistance with finding a clinical trial they are eligible for."** Given the degree to which patients and caregivers rely on medical providers for information and advice around clinical trials, it is important for providers to have this information available and accessible in a variety of formats, particularly for older and low-literacy populations.

- As a first step, MDA will be including clinical trial information in its **Welcome Packets** that can be used as a conversation starter between patients, caregivers, and health care providers.
- MDA's **clinical trial finder tool**, available through the MDA website, contains a series of questions designed to help patients and caregivers identify trials that may be a good match. However, currently less than a third (31%) of the community is familiar with this tool.
- MDA also disseminates information about clinical trials to the neuromuscular community through its website as well as e-mails, blogs, social media, and Quest magazine.
- Medical and genetic data from MDA's **MOVR** (neuroMuscular ObserVational Research) Data Hub enables health care professionals to track or measure a number of health-related or quality-of-life outcomes in individuals with a specific disease or condition and can help doctors quickly identify patients who may benefit from new therapies or who may want to participate in a clinical trial. Currently four diseases are included in MOVR: ALS, SMA, DMD, and BMD. MDA plans to include the addition of LGMD, FSHD, and Pompe in 2019.
- MDA is also committed to developing more clinical-trial-ready sites and supports multiple clinical research networks focused on specific diseases under the MDA umbrella. Each consists of networks of multiple MDA Care Centers with special expertise in clinical research. The Care Centers work together on projects selected by the network, such as natural history studies, endpoint determinations, and other clinical research efforts. The investigators of the networks meet regularly by phone and in person to discuss ongoing and novel programs. Further, involvement in clinical trials and educating patients about where to find information about clinical trials are incorporated into the MDA Care Center criteria.

Finding 4: Mobility and independence are top of mind for the neuromuscular disease community.

Independence and mobility are a major concern for most (64%) of the community, topping the list of concerns for both adult patients and caregivers. In general, concerns around independence cut across demographics (income, education, race/ethnicity), however, adult patients in their 50s and 60s, as well as those diagnosed with LGMD and SMA, were slightly more likely to list independence as a major concern.

While independence is a top concern for adult patients, the population is still empowered and positive in their outlook. 7 in 10 (70%) adult patients agree with the statement "I/my loved one can lead an independent, productive life with a neuromuscular disease." Adult patients under 50, those diagnosed more than 10 years ago, and patients with CMT, SMA, and BMD tend to be the most optimistic.

Figure 17: Independence regarding mobility is a major concern.

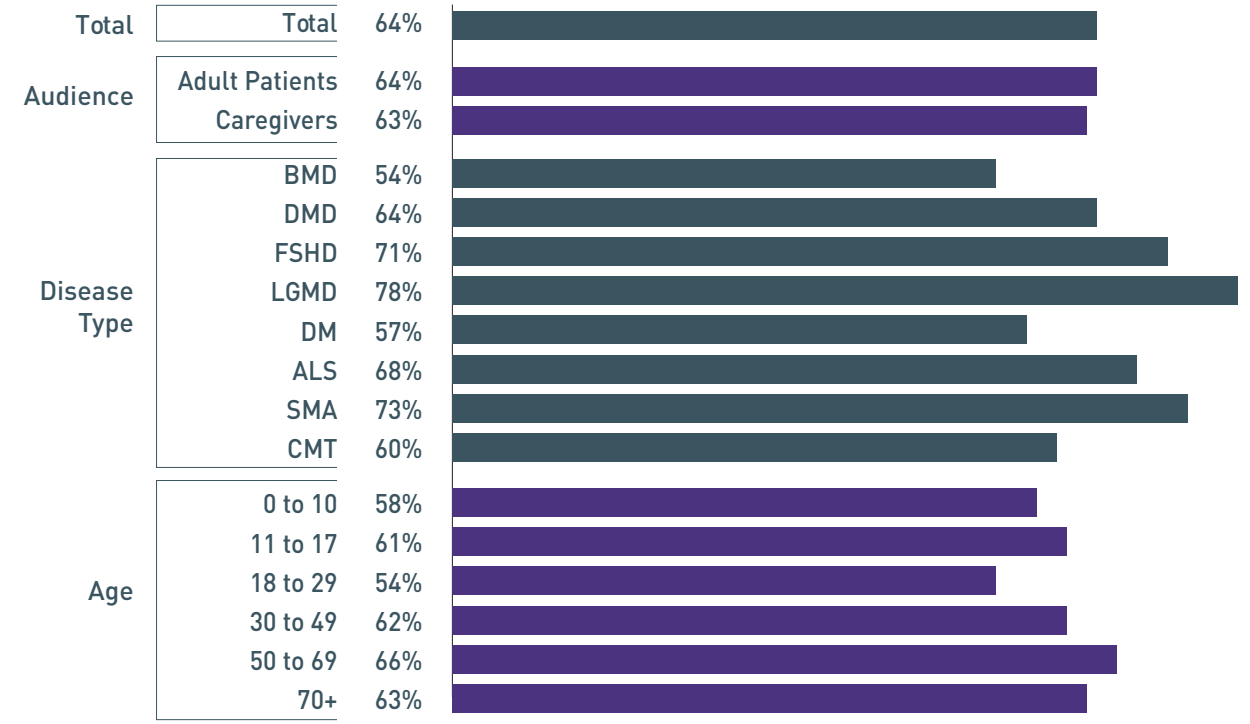
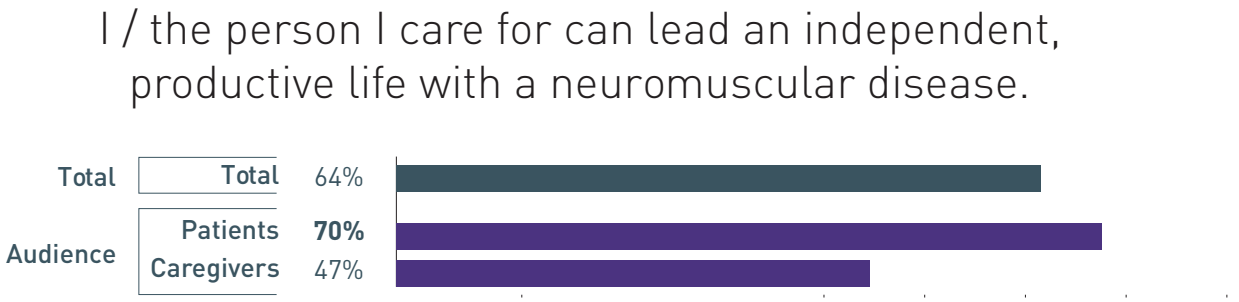


Figure 18: Adult patients are more optimistic about their independence than caregivers. (Bolding indicates statistical significance.)



This sentiment, however, is much lower for caregivers – just 47 percent agree with this statement. Parents are slightly more optimistic about independence than spouses (53% of spouses v. 44% of parents disagree with the statement). Open-ended responses reveal that this sentiment may stem from overall feelings of uncertainty about the future (which tops the list of major concerns for caregivers) as well as the emotional and physical impacts of caregiving.

A strong desire for independence translates into high interest in services and resources that foster independence: 73% say they are interested in "information and resources for independent living (e.g. finding, managing, and paying for personal care, accessible housing, financial management, etc.)" and, among adult patients, "ways to increase independence" ranks third out of 23 different topics related to living with neuromuscular disease. Respondents who listed independence as a major concern were also significantly more likely to have major concerns about access to lifestyle services including disability-modified vehicles, accessible housing, long-term planning and career goals, and air travel.

Caregivers' comments shed light on their own challenges

"I'm afraid of the future. Every day my son seems to get worse, is in more pain, and his legs are weaker."
-Caregiver/parent

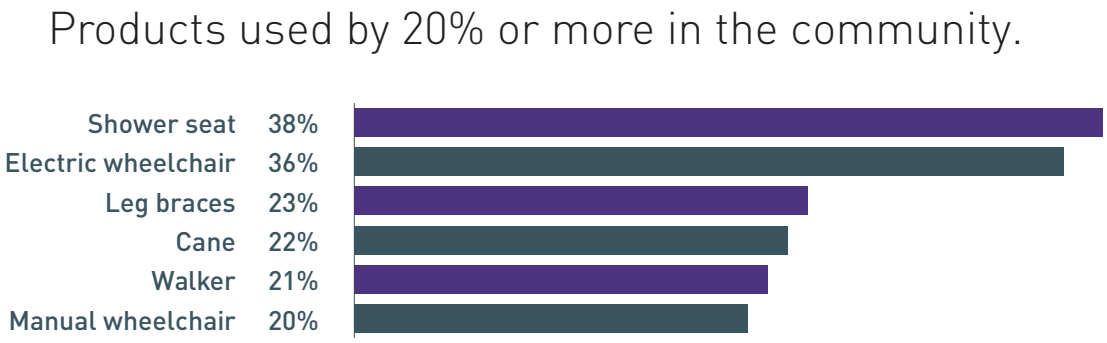
"Feeling alone…wanting my spouse to have their independence yet sometimes they do or not do some things they could or should do."
-Caregiver/spouse

"Watching him suffer and decline is horrible."
-Caregiver/spouse

"My biggest challenge is finding the energy to care for my son 24 hours a day, 7 days a week."
– Caregiver/parent

The community relies on a range of products/services to aid in their mobility. Topping the list are equipment that enables ambulation, including electric wheelchairs (36%), leg braces (23%), canes (22%), walkers (21%), and manual wheelchairs (20%). Moreover, the community is interacting frequently with equipment and other goods/services providers, including standard durable and customized medical equipment providers, pharmacies, and online stores like Amazon for assistance with their daily needs. These providers offer an important opportunity for patients and caregivers to access the equipment and support they need for managing their disease and living more independently.

Figure 19. Top-mentioned products and providers of goods and services among neuromuscular community.



Solutions: Online Ordering and Deliveries Are a Game Changer

Online ordering and delivery of groceries and other goods and supplies through providers such as Amazon are widely available and utilized by the community. In fact, almost 4 in 10 (38%) mention Amazon as helping with their day-to-day needs. This technology enables individuals living with neuromuscular disease to become more self-sufficient.

Finding 5: MDA is a go-to resource.

The neuromuscular disease community is eager for information and counts on MDA to fill this need. Most of those who responded to this survey (75%) say they use MDA for information on issues related to neuromuscular disease, followed to a lesser extent by health care providers (66%), internet or Google searches (52%), and disease-specific organizations (42%).

The MDA website is a key touchpoint for the neuromuscular community, with more than 70% saying they have visited the site. Over half (56%) say have reached out in other ways to MDA for information/assistance.

More than MDA services garner high interest from the community. Among both patients and caregivers, the MDA Resource Center staffed by social workers and employees with significant experience with neuromuscular disease tops the list, followed by specialized medical care, help finding clinical trials, and assistance with independent living. However, notable gaps exist between interest in and awareness of these currently offered services – highlighting an important need to promote greater awareness of these in-demand resources. The Resource Center is working to create more user-friendly access to support information through smart technology and by promoting the center more broadly.

Figure 20: Information sources for the neuromuscular disease community. (Bolding indicates statistical significance.)

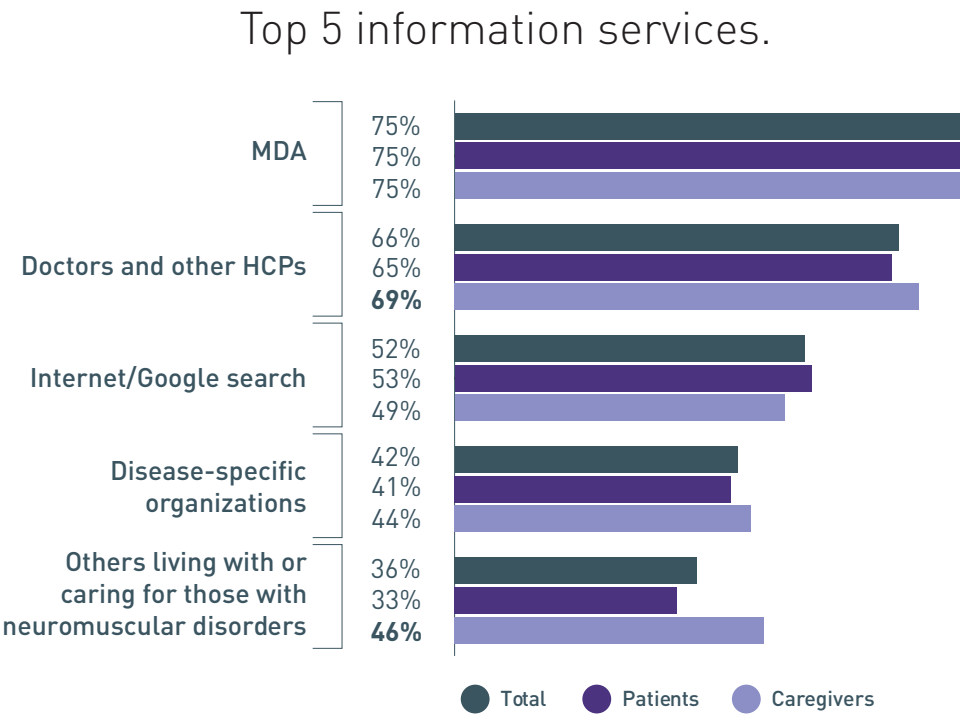
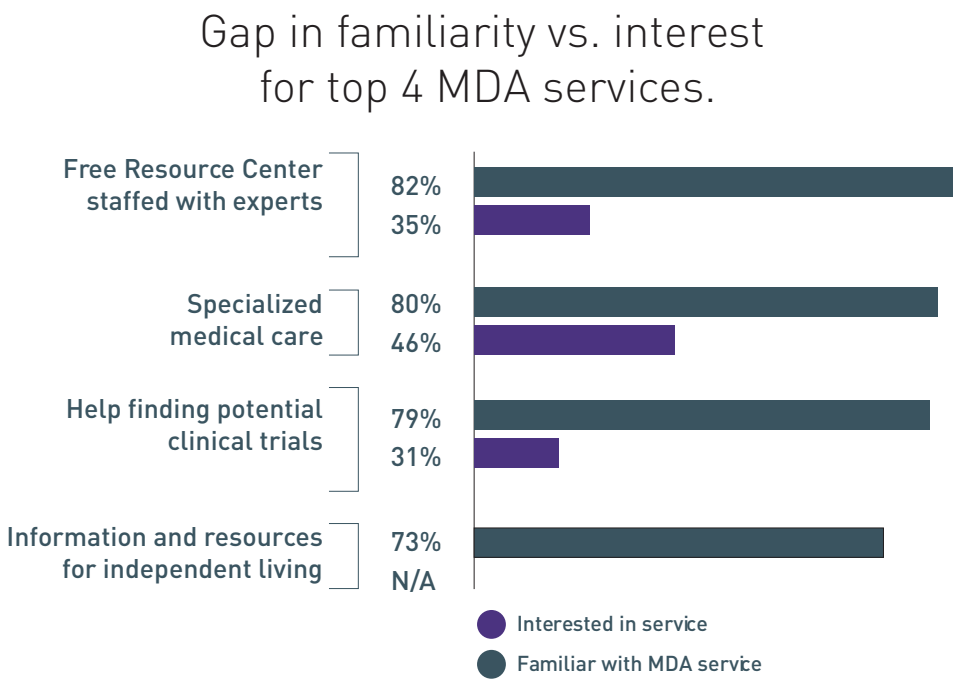


Figure 21: More need information about MDA's in-demand services



More vulnerable patient populations are a critical audience for MDA's resources and support services. Statistical analysis was used to sort adult patients into segments based on a different set of characteristics, attitudes, and needs when it comes to living with neuromuscular disease. There is a segment of the adult patient community that emerges as the most vulnerable: They are more likely to be in poor physical and mental health, and more disenfranchised economically. This group is more concerned about the range of challenges facing people with neuromuscular disease and highly interested in services and support from MDA. They, more so than others, long for connection: 43% say connecting with other individuals and caregivers living with neuromuscular disease is extremely/very important, and express strong interest in virtual/online and in-person forums that enable increased interaction with others in the community.

With greater outreach and targeting of its current resources and services, MDA is well-equipped to meet many of the most pressing needs of the community generally, and the most vulnerable patient populations particularly.

IMPLICATIONS

This survey is the first widespread study about the experiences, challenges, and needs of individuals and families living with neuromuscular disease. It begins to answer many pressing questions and yields valuable insights that can inform the work of MDA and others serving the neuromuscular community. But it brings up just as many questions and ideas on how the field can do more.

For MDA, this study marks the beginning of a continuing process to hear the voice of the community, to gather input and to ensure their needs are being met. As MDA continues to explore these issues, subsequent efforts will drill further into other areas. For instance, knowing more about the dynamics between patients and their health care providers could help to improve communication around such important topics as genetic testing and clinical trials. By continuing to keep its finger on the pulse of the community, MDA hopes to ensure that all members are heard and their needs are addressed.

To help achieve these goals, MDA is calling on the entire neuromuscular community to join us in our advocacy efforts in the coming year. Our work to improve access to care, bolster early diagnosis and intervention through newborn screening, advance genetic testing, and increase access to travel will be reflective of the results of this survey. We are committed to fostering and promoting change that will make a difference to the community we serve, but we are only as strong as the commitment of our advocates. Join us here: www.mda.org/advocacy.

METHODOLOGY

Edge Research conducted an online survey of 3,362 members of the MDA community. This included 2,430 adult patients and 932 parents/caregivers (727 parents; 187 spouse/partners; 18 other). The survey was in the field from January 18 through February 20, 2018, and was distributed by MDA through multiple channels to encourage participation: emails from MDA and partner organizations, the MDA website, and social media.

The survey instrument was peer reviewed by MDA staff and other experts in the field and was pretested with several patients and caregivers for comprehension and usability.

The survey consisted of more than 50 questions covering topics about experiences and challenges around disease management, treatment burdens and trade-offs, sources of information and support, awareness and interest in MDA resources and services, and demographics. The survey included a few open-format questions where participants could type in their feedback. Those are represented in the verbatim quotes throughout the report.

As with all online surveys, this survey uses a convenience sample of respondents drawn from multiple channels. As such, findings are not generalizable to the neuromuscular population as a whole.

Contributors:

Presented by MDA

MDA is committed to transforming the lives of people affected by muscular dystrophy, ALS, and related neuromuscular diseases. We do this through innovations in science and innovations in care. As the largest source of funding for neuromuscular disease research outside of the federal government, MDA has committed more than $1 billion since our inception to accelerate the discovery of therapies and cures. Research we have supported is directly linked to life-changing therapies across multiple neuromuscular diseases. MDA's MOVR is the first and only data hub that aggregates clinical, genetic, and patient-reported data for multiple neuromuscular diseases to improve health outcomes and accelerate drug development. MDA supports the largest network of multidisciplinary clinics providing best-in-class care at more than 150 of the nation's top medical institutions, and our Resource Center serves the community with one-on-one specialized support. We offer educational conferences, events, and materials for families and health care providers. Each year thousands of children and young adults learn vital life skills and gain independence at MDA Summer Camp and through recreational programs, at no cost to families. For more information, visit mda.org

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