



Needs management in families affected by childhood-onset dystrophinopathies

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Abstract

Purpose: To collect information about the needs of families affected by childhood-onset dystrophinopathies residing in the United States.

Methods: Individuals with an eligible dystrophinopathy were identified by the Muscular Dystrophy Surveillance, Tracking, and Research network. Between September 2008 and December 2012, 272 caregivers completed a 48-item survey about needs related to information, healthcare services, psychosocial issues, finances, caregiver demographics, and the individual's functioning.

Results: Overall, at least 80% of the survey items were identified as needs for more than one-half of caregivers. Among the needs identified, physical health and access to information were currently managed for most caregivers. Items identified as needed but managed less consistently were funding for needs not covered by insurance and psychosocial support.

Conclusions: Healthcare providers, public health practitioners, and policymakers should be aware of the many needs reported by caregivers, and focus on addressing gaps in provision of needed financial and psychosocial services.

Keywords

Becker muscular dystrophy, Duchenne muscular dystrophy, dystrophinopathy, caregivers, needs assessment, survey

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Duchenne muscular dystrophy (DMD) and Becker muscular dystrophy (BMD), herein referred to as dystrophinopathies, are allelic X-linked disorders caused by a mutation in the *DMD* gene.¹ Currently, there are no curative treatments. Oral corticosteroids are associated with improved outcomes, including prolonged walking and delayed orthopedic, cardiac, and respiratory complications in DMD.^{2–16} In addition, monitoring and intervention on aspects of cardio-pulmonary manifestations has likely improved longevity.^{17,18} More recently, use of medications, such as Eteplirsen, may modify disease progression in a subset of males with specific *DMD* mutations amenable to exon 51 skipping.^{2–15}

Although available treatments modify disease course, declines in muscle integrity and multi-system functional deterioration continue to be significant problems requiring long-term management.^{19–21} Consequently, family needs associated with a dystrophinopathy diagnosis remain and may increase as diagnosed individuals survive longer.²⁰ Studies on the quality of life of individuals diagnosed with a dystrophinopathy and on the burden to their families have

reported lower adaptability among those individuals with increasing health demands.^{22–31} Although the measurement of burden and adaptation have varied across studies, adequate practical and psychological resources have been shown to reduce the effect of these demands on the diagnosed individual and their families.^{23,26,27,32,33}

To assist healthcare providers, public health practitioners, and policymakers in gaining a better understanding of the needs of caregivers and families of individuals with a dystrophinopathy diagnosis, we administered a comprehensive

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survey to caregivers of individuals with a childhood-onset dystrophinopathy identified from a multi-site, population-based cohort. The purposes of our survey were to describe the needs of caregivers and diagnosed individuals, the association of needs with stages of disease, and family demographics, and to identify gaps in care.

Methods

The Muscular Dystrophy Surveillance, Tracking, and Research Network (MD STARnet) is a retrospective surveillance cohort of eligible individuals diagnosed with a childhood-onset dystrophinopathy. MD STARnet methods have been described elsewhere.^{34,35} Briefly, starting in 2004, the MD STARnet retrospectively ascertained individuals with childhood-onset dystrophinopathy who were born since 1 January 1982, diagnosed by age 21 years, and resided in an MD STARnet site (Arizona (AZ), Colorado (CO), Iowa (IA), and the western 12 counties of New York State [wNY]). Georgia (GA) and Hawaii (HI) joined the MD STARnet in 2005 and 2008, respectively. Since the initiation of surveillance, medical record abstraction was completed annually for all retrospectively ascertained individuals and newly diagnosed individuals through 31 December 2011 (for individuals identified before September 2011), 31 December 2012 (for individuals identified after September 2011), or until death or migration out of an MD STARnet site. Trained medical abstractors reviewed medical records and entered data into an electronic surveillance application. Abstracted data included sociodemographic data, clinical signs and symptoms, diagnostic/clinical tests, family history of dystrophinopathy, medical treatments received, and medical complications experienced. Data on schooling, rehabilitation, mobility, and neurobehavioral problems were also collected.

Needs assessment survey

The survey was developed by the MD STARnet Needs Assessment workgroup, comprising representatives from each MD STARnet site. The survey expanded on an initial needs assessment funded by the Centers for Disease Control and Prevention (CDC) and piloted in Iowa in 2005–2006. Questions included caregiver sociodemographic information (marital status (single, never married; married or living as married; widowed; separated; divorced), race (American Indian or Alaska Native; Asian; Black or African American; Native Hawaiian or Other Pacific Islander; White), ethnicity (Hispanic or Latino; Not Hispanic or Latino), education (grade school; junior high; high school, no diploma; high school with diploma; some college, no diploma; technical college or associate degree; bachelor's degree; graduate or professional degree), and annual household income (<US\$10,000; US\$10,000–US\$19,999; US\$20,000–US\$29,999; US\$30,000–US\$39,999; US\$40,000–

US\$49,999; US\$50,000–US\$59,999; US\$60,000–US\$69,999; US\$70,000–US\$79,999; US\$80,000–US\$89,999; US\$90,000–US\$99,999; [>US\$100,000]), household composition (number), physical disabilities of family members in the household (yes/no), caregiving provided by family members (yes/no), with whom the child resided (both parents, same household; one parent; both parents, different households; relative; foster home; care facility or group home; independently; spouse or partner; other), current ambulation (walking without help; walking with help; manual wheelchair, full-time; power scooter, full-time; power wheelchair, full-time; cannot operate power mobility) and respiratory function (no assistance; bi-pap or c-pap with sleep; daytime bi-pap or c-pap; bi-pap or c-pap with sleep and sip-n-puff mouthpiece, breath-stacking during the day; tracheostomy with part-time ventilation; tracheostomy with full-time ventilation), assistance with activities of daily living (yes/no), and health-related services received (available and helpful; frequency).

The survey consisted of 48 items across 5 domains: home, community, and durable equipment (13 items); healthcare management of common morbidities (10 items); access to factual information about genetic testing, research, and planning (11 items); psychosocial issues for the individual (5 items); and psychosocial issues for the parent/caregiver and family (9 items). These items were chosen based on a literature review, clinical experience, and pilot data that included the opportunity for families to list needs not included among the item responses in the pilot survey. Each need was scored for management by the caregiver as “Currently being managed or has usually been managed,” “Is not currently or has not usually been managed,” and “Does not apply.” Finally, caregivers were asked to select their three highest unmanaged needs items from the 48 items listed.

Survey sample and data collection

The eligible sample comprised 520 primary caregivers of living individuals diagnosed with a dystrophinopathy, who had either a positive DNA or muscle biopsy test result or a positive family history of a dystrophinopathy and were identified from surveillance years 2004–2011 (see Figure 1). Among multiple-affected families, caregivers were asked to report on their oldest affected son. Surveys were administered from January 2008 through August 2012. Recruitment was initiated by mail either directly to the eligible caregiver (CO, HI, IA, NY, or GA) or through the primary healthcare provider (AZ). Following a pre-contact letter informing the caregiver about the study, an introductory packet—consisting of a detailed explanation of the study, a project brochure, information on frequently asked questions and rights of research subjects, US\$20 compensation, and the survey—was mailed. A systematic follow-up procedure included reminder telephone calls every 2 weeks followed by a mailed reminder letter if contact could

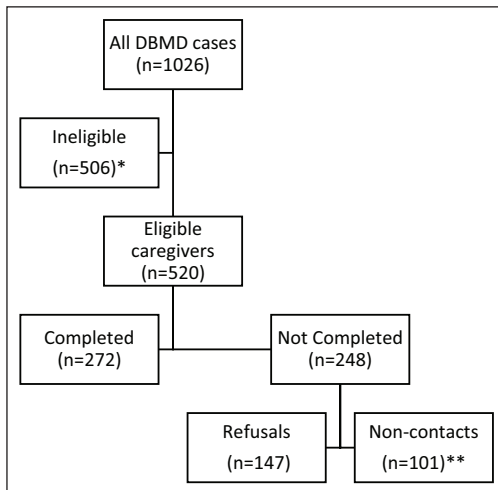


Figure 1. Flowchart of eligible caregivers and completed questionnaires.

*Ineligible caregivers included those of deceased males or those of males with possible or asymptomatic diagnoses, or affected females.

**Non-contacts include mailings to an unverified address that were not verified, returned mail with undeliverable address, and unverifiable address for mailings.

not be made by telephone, until the survey was returned, or a maximum of three unsuccessful follow-up attempts were completed. The final reminder mailing included an additional copy of the survey. The survey was available in English or Spanish and typically took 30 min to complete. Returned surveys were evaluated for completeness; caregivers were contacted if missing data were identified. Surveys were scanned using Teleform (Verity, Inc. Sunnyvale, CA, version 10.6, 2012), and scanned data were reviewed manually for accuracy by two reviewers. Local institutional review board approval to conduct survey data collection with caregivers was obtained at each MD STARnet site. Consent to participate was received by signature (CO, wNY) or implied through completion of the survey (AZ, GA, IA).

Statistical analysis

Item response categories were recoded in two steps in order to identify needs and the management of those needs. First, responses were recoded to reflect the presence of a need based on current or past management of an item (Not needed [0] = “Does not apply”; Needed [1] = “Currently being managed or has usually been managed” OR “Is not currently or has not usually been managed”). Missing items were excluded when recoding presence of needs. Second, a variable representing management of needs was created by recoding only those items for which management was reported (Not managed [0] = “Is not currently or has not usually been managed”; Managed [1] = “Currently being managed or has usually been managed”). “Does not apply” and missing responses were excluded when recoding management of needs.

Using available surveillance data, we compared selected characteristics for all eligible caregivers and those who responded to the survey. Analyses included item frequency counts for identified needs and reported management of each need. Chi-square analyses examined associations between identified needs and management of needs and ambulation status of the individual (ambulatory; non-ambulatory) and caregiver demographics (annual income [$<$ US\$30,000; \geq US\$30,000] and race/ethnicity (non-Hispanic white; minority race/ethnicity)). A p -value < 0.05 was used to determine statistically significant associations.

Results

The American Association for Public Opinion Research (AAPOR) calculator was used to estimate outcome rates; formulas that estimated the number of potentially eligible participants from the number of participants with undetermined eligibility were used.³⁶ Survey responses from HI ($n=2$) were excluded from analyses due to the limited follow-up conducted with primary caregivers. For the remaining sites, the response rate (number of completed surveys by caregivers divided by the number of eligible caregivers in the sample) was 59%. The contact rate (proportion of households in which the caregiver was reached) was 85%. The cooperation rate (proportion of all completed surveys from all caregivers where contact was verified) was 69%.

Compared to all eligible caregivers in the MD STARnet cohort, caregivers who completed the survey were more likely to be non-Hispanic White and have higher education than those who did not respond (data not shown). No other characteristics differed statistically between all eligible caregivers and those who responded to the survey. At the time of survey completion, the average age of the individual with DMD was 14.8 years ($SD=6.9$); 56% were non-ambulatory, and 29% required respiratory assistance (Supplemental Table 1). Among caregivers, 78% were married, 50% had some college or higher, 62% had an annual income of or above US \$30,000 with an average of four persons within a household supported by this income, and 15% had other persons in the household affected by a disability. For the affected individual, over 70% lived with both parents in the same household, over 50% used a wheelchair full-time, and nearly one-third required respiratory intervention. Over 90% of caregivers provided personal and health care functions for their child and nearly 70% paid others to assist.

Needs of families

The top 10 needs identified are shown by rank order in Table 1 together with the percentage of respondents who affirmed that this was a need (e.g. “Currently being managed or has usually been managed” OR “Is not currently or has not usually been managed”). Tables 2–4 display identified needs related to ambulation status, household income, and

caregiver race/ethnicity. Overall, the top 10 needs by these subgroups were similar to those identified by the overall sample, although the ranking differed and different subgroups identified some unique needs (highlighted in tables).

Descriptive statistics for needs, as well as results for comparisons by individual ambulation status, and caregiver reported annual household income and race/ethnicity are presented in Supplemental Table 2. As expected, given the development of the survey, 80% or more of the survey items

Table 1. Top 10 needs by all caregivers, the Muscular Dystrophy Surveillance, Research, and Tracking network, 2008–2012 (n = 272).

Needs	% needed
Information on MDA, PPMD or similar organizations	96.3
Access to research updates	95.9
Information about the course of the disease	95.5
Access to research participation	95.2
Information about financial resources (private, state, federal)	94.1
Information for talking to family/friends about health concerns	92.6
Balancing work, family, and caregiving	91.1
Finding time and energy for yourself	89.9
Finding time and energy for adult relationships	88.8
Suggestions for recreational activities alone or with family/friends	88.4

MDA: muscular dystrophy association; PPMD: parent project muscular dystrophy.
Needs were recoded as not needed = 0 or needed = 1; thus, the percentage reported in the table represents the percentage of caregivers that identified an item as needed.

Table 2. Top 10 needs by ambulation status, the Muscular Dystrophy Surveillance, Research, and Tracking network, 2008–2012 (n = 272).

Caregivers of non-ambulatory individuals	% needed	Caregivers of ambulatory individuals	% needed
Completing personal care activities (bathing, lifting, toileting)	98.0	Access to research updates	96.5
Dental care	97.3	Information on MDA, PPMD or similar organizations	95.6
Information on MDA, PPMD or similar organizations	96.7	Information about the course of the disease	93.9
Information about financial resources (private, state, federal)	96.7	Access to research participation	93.8
Information about the course of the disease	96.6	Information for talking to family/friends about health concerns	93
Access to research participation	96.0	Information about financial resources (private, state, federal)	90.4
Balancing work, family, and caregiving	96.0	Information about inheritance and carrier testing	86.7
Getting funding/insurance to pay for equipment	96.0	Balancing work, family, and caregiving	83.9
Access to research updates	95.4	Finding time and energy for yourself	82.1
Finding time and energy for yourself	98.0	Finding time and energy for adult relationships	82.1

MDA: muscular dystrophy association; PPMD: parent project muscular dystrophy.
Needs were recoded as not needed = 0 or needed = 1; thus, the percentage reported in the table represents the percentage of caregivers that identified an item as needed. Gray cells identify unique needs across ambulation status. Needs are presented from highest to lowest order based on percentages of caregivers who reported the item as needed.

were identified as needs by more than 50% of all responding caregivers (Supplemental Table 2). Overall, 33 of the 48 queried needs were significantly different by ambulation status with needs more common among caregivers of non-ambulatory individuals.

Managing needs of families

The top 10 unmanaged needs (e.g. “Is not currently or has not usually been managed”) for all respondents that identified an item as a need are shown in Table 5. The majority of the unmet needs were in the realms of information, psychosocial support, and access to funding. Consistent with the data on identified needs, the unmet needs in subgroups (shown in Tables 6–8) were very similar, with a few unique unmet needs in each subgroup and differences in rank order.

Descriptive statistics for management of needs, as well as results for comparisons by individual ambulation status, caregiver reported annual income and race/ethnicity are presented in Supplemental Table 3. Relatively few significant differences were found for comparisons of subgroups regarding management of needs. Only 12 of 48 comparisons were differed significantly by ambulation status and those were equally distributed across all domains. Similarly, the management of nine needs differed by annual income, with poorer management of home and vehicle modifications, and time and energy and relationships among household with annual incomes <US\$30,000 compared to those with higher incomes. Only four comparisons were significant by race/ethnicity; poorer management of funding needs for home and vehicle modifications, and

Table 3. Top 10 needs by caregiver reported annual household income, the Muscular Dystrophy Surveillance, Research, and Tracking network, 2008–2012 (n = 272).

<US\$30,000 household income	% Needed	≥US\$30,000 household income	% needed
Information on MDA, PPMD or similar organizations	94.9	Information on MDA, PPMD or similar organizations	97.0
Access to research updates	93.8	Access to research updates	97.0
Information about financial resources (private, state, federal)	93.8	Information about the course of the disease	96.4
Information about the course of the disease	93.7	Access to research participation	96.4
Access to research participation	91.3	Information for talking to family/friends about health concerns	95.2
Dental care	91.1	Information about financial resources (private, state, federal)	94.7
Joint contractures and muscle weakness	91.0	Balancing work, family, and caregiving	92.3
Suggestions for recreational activities alone or with family/friends	89.6	Finding time and energy for yourself	91.6
Information for talking to family/friends about health concerns	88.8	Finding time and energy for adult relationships	91.0
Balancing work, family, and caregiving	88.6	Making and maintaining age-appropriate friendships	89.7

MDA: muscular dystrophy association; PPMD: parent project muscular dystrophy.

Needs were recoded as not needed = 0 or needed = 1; thus, the percentage reported in the table represents the percentage of caregivers that identified an item as needed. Gray cells identify unique needs across household income. Needs are presented from highest to lowest order based on percentages of caregivers who reported the item as needed.

Table 4. Top 10 needs by caregiver race/ethnicity, the Muscular Dystrophy Surveillance, Research, and Tracking network, 2008–2012 (n = 272).

Minority caregivers	% needed	Non-minority caregivers	% needed
Information on MDA, PPMD or similar organizations	94.8	Information on MDA, PPMD or similar organizations	97.6
Access to research updates	91.4	Access to research updates	97.6
Balancing work, family, and caregiving	91.4	Information about the course of the disease	97.6
Dental care	91.4	Access to research participation	96.6
Access to research participation	91.2	Information about financial resources (private, state, federal)	96.2
Information for talking to family/friends about health concerns	91.2	Information for talking to family/friends about health concerns	93.8
Information about the course of the disease	91.1	Balancing work, family, and caregiving	91.8
Information about financial resources (private, state, federal)	89.7	Finding time and energy for yourself	91.3
Suggestions for recreational activities alone or with family/friends	89.3	Finding time and energy for adult relationships	90.8
Finding time and energy for yourself	87.9	Making and maintaining age-appropriate friendships	89.2

MDA: muscular dystrophy association; PPMD: parent project muscular dystrophy.

Needs were recoded as not needed = 0 or needed = 1; thus, the percentage reported in the table represents the percentage of caregivers that identified an item as needed. Gray cells identify unique needs across race/ethnicity. Needs are presented from highest to lowest order based on percentages of caregivers who reported the item as needed.

access to information was found among minority caregivers compared to non-minority.

Most unmanaged needs

In a further effort to identify high-priority needs, caregivers were asked to select the top 3 issues from the list of 48

survey items that they felt had been the most difficult to deal with or were (usually) not managed. Table 9 shows the items identified by responders, together with the percentage of caregivers who chose that item. Consistent with the overall analysis, unmanaged needs most commonly listed were related to the realms of financial and psychosocial supports.

Table 5. Top 10 unmanaged needs for all caregivers, the Muscular Dystrophy Surveillance, Research, and Tracking network, 2008–2012 (n = 272).

Needs	% managed
Information about jobs/future planning for the male with DBMD	29.7
Information about transitioning to independent adult life	32.5
Finding funding for vehicle modifications	37.3
Counseling to help emotional adjustment of parent (depression, fear, loneliness)	40.1
Arranging or getting caregiver access to marriage or relationship counseling	41.1
Finding funding for home modifications	44.3
Information about how to prepare advance directives: living wills and designating durable power of attorney for healthcare	45.4
Finding or attending parent, spouse or sibling support groups	46.8
Information about financial resources (private, state, federal)	47.1
Counseling to help emotional adjustment of individual with DMD (depression, fear, loneliness)	49.3

DBMD: Duchenne/Becker muscular dystrophy; DMD: Duchenne muscular dystrophy.

Management of needs was recoded as not managed = 0 or managed = 1; thus, the percentage reported in the table represents the percentage of caregivers that identified a need as managed.

Table 6. Top 10 unmanaged needs by ambulation status, the Muscular Dystrophy Surveillance, Research, and Tracking network, 2008–2012 (n = 272).

Caregivers of non-ambulatory individuals	% managed	Caregivers of ambulatory individuals	% managed
Information about jobs/future planning for individual with DBMD	32.8	Finding funding for vehicle modifications	16.7
Information about transitioning to independent adult life	34.8	Information about jobs/future planning for individual with DBMD	23.5
Arranging or getting access to marriage or relationship counseling	37.3	Information about transitioning to independent adult life	25.9
Caregiver counseling for emotional adjustment (depression, fear, loneliness)	39.8	Information about how to prepare advance directives: living wills and designating durable power of attorney for healthcare	38.8
Finding funding for vehicle modifications	43.0	Finding funding for home modifications	42.0
Finding or attending parent, spouse or sibling support groups	43.8	Caregiver counseling for emotional adjustment (depression, fear, loneliness)	42.4
Finding funding for home modifications	44.5	Finding a builder/contractor for home modifications	43.6
Information about how to prepare advance directives: living wills and designating durable power of attorney for healthcare	48.5	Information about financial resources (private, state, federal)	44.7
Information about financial resources (private, state, federal)	48.6	Arranging or getting access to marriage or relationship counseling	46.7
Affected individual counseling for emotional adjustment (depression, fear, loneliness)	48.7	Finding or attending parent, spouse or sibling support groups	48.7

DBMD: Duchenne/Becker muscular dystrophy.

Management of needs was recoded as not managed = 0 or managed = 1; thus, the percentage reported in the table represents the percentage of caregivers that identified a need as managed. Gray cells identify unique unmanaged needs across ambulation status. Unmanaged needs were selected based on percentage of caregivers reporting management of the need, for example, only 16.7% of caregivers of ambulatory males report finding funding for vehicle modifications as managed.

Discussion

Our study systematically identified needs and management of these needs by caregivers and families of individuals diagnosed with a childhood-onset dystrophinopathy across a range of domains. The most commonly identified needs included access to information about support groups, research, disease progression, and balancing time and energy. Overall, although we found that families have many

needs, caregivers responded that most medical and information needs were being managed. Gaps in meeting individual and family needs remained, however, primarily in the spheres of availability of information about future planning and transitions, financial assistance, and psychosocial supports for family members. Financial-related needs were unmanaged for over one-half of caregivers and were the most frequently selected unmanaged needs from all available items.

Table 7. Top 10 unmanaged needs by caregiver reported annual household income, the Muscular Dystrophy Surveillance, Research, and Tracking network, 2008–2012 (n = 272).

<US\$30,000 Household income	% managed	≥US\$30,000 Household income	% managed
Arranging or getting access to marriage or relationship counseling	22.9	Information about transitioning to independent adult life	30.6
Finding funding for vehicle modifications	27.6	Information about jobs/future planning for individual with DBMD	30.7
Information about jobs/future planning for individual with DBMD	29.6	Caregiver counseling for emotional adjustment (depression, fear, loneliness)	42.0
Finding or attending parent, spouse or sibling support groups	32.1	Finding funding for vehicle modifications	43.7
Finding funding for home modifications	33.9	Information about financial resources (private, state, federal)	45.6
Information about transitioning to independent adult life	34.0	Information about how to prepare advance directives: living wills and designating durable power of attorney for healthcare	49.6
Caregiver counseling for emotional adjustment (depression, fear, loneliness)	35.3	Arranging or getting access to marriage or relationship counseling	50.0
Information about how to prepare advance directives: living wills and designating durable power of attorney for healthcare	36.1	Finding or attending parent, spouse or sibling support groups	51.1
Finding a builder/contractor for home modifications	37.5	Finding funding for home modifications	51.7
Affected individual counseling for emotional adjustment (depression, fear, loneliness)	38.9	Affected individual counseling for emotional adjustment (depression, fear, loneliness)	54.3

DBMD: Duchenne/Becker muscular dystrophy.

Management of needs was recoded as not managed = 0 or managed = 1; thus, the percentage reported in the table represents the percentage of caregivers that identified a need as managed. Gray cells identify unique unmanaged needs across household income. Unmanaged needs were selected based on percentage of caregivers reporting management of the need, for example, only 22.9% of caregivers from lower income households report getting access to marriage counseling as managed.

Table 8. Top 10 unmanaged needs by caregiver race/ethnicity, the Muscular Dystrophy Surveillance, Research, and Tracking network, 2008–2012 (n = 272).

Minority caregivers	% managed	Non-minority caregivers	% managed
Finding funding for vehicle modifications	20.9	Information about jobs/future planning for the individual with DBMD	28.5
Information about transitioning to independent adult life	32.5	Information about transitioning to independent adult life	32.0
Information about jobs/future planning for individual with DBMD	33.3	Caregiver counseling for emotional adjustment (depression, fear, loneliness)	40.7
Finding a builder/contractor for home modifications	36.8	Arranging or getting access to marriage or relationship counseling	41.7
Arranging or getting access to marriage or relationship counseling	37.0	Finding funding for vehicle modifications	42.4
Finding funding for home modifications	37.8	Finding funding for home modifications	46.3
Caregiver counseling for emotional adjustment (depression, fear, loneliness)	38.9	Information about how to prepare advance directives: living wills and designating durable power of attorney for healthcare	46.6
Information about how to prepare advance directives: living wills and designating durable power of attorney for healthcare	39.5	Finding or attending parent, spouse or sibling support groups	47.1
Affected individual counseling for emotional adjustment (depression, fear, loneliness)	42.5	Information about financial resources (private, state, federal)	47.3
Patient discussion/support groups	43.2	Affected individual counseling for emotional adjustment (depression, fear, loneliness)	51.6

DBMD: Duchenne/Becker muscular dystrophy.

Management of needs was recoded as not managed = 0 or managed = 1; thus, the percentage reported in the table represents the percentage of caregivers that identified a need as managed. Gray cells identify unique unmanaged needs across caregiver race/ethnicity. Unmanaged needs were selected based on percentage of caregivers reporting management of the need, for example, only 22.9% of caregivers from lower income households report getting access to marriage counseling as managed.

Table 9. Items listed as most unmanaged needs by caregivers, the Muscular Dystrophy Surveillance, Research, and Tracking network, 2008–2012 (n = 272).

Most unmanaged needs	% of caregivers
Information about financial resources (private, state, federal, etc.)	19.0
Finding funding for home modifications	16.8
Counseling to help emotional adjustment of affected male	16.1
Finding funding for vehicle modifications	15.0
Information about jobs/future planning for the male with DBMD	13.1
Finding time and energy for yourself (personal time for the caregiver)	12.4
Making and maintaining age-appropriate friendships for the male with DBMD	10.9
Finding funding and available respite care so caregivers can be gone	9.8
Finding time and energy for adult relationships (spouse/significant other)	9.5

DBMD: Duchenne/Becker muscular dystrophy.

Our findings are in keeping with others who have documented the financial burden of DMD. Survey data from caregivers of individuals with DMD followed in Muscular Dystrophy Association (MDA) clinics reported costs for durable medical equipment are, on average, approximately US\$1100, modifying motor vehicle US\$1600, and home modification US\$3050.²⁹ Similarly, a Treat NeuroMuscular Disease (TREAT-NMD) DMD registry survey of caregivers residing in the United States reported total annual costs of approximately US\$7900 for aids and devices and investments in home and vehicle modifications.²⁸ Several analyses have reported that the healthcare costs for an individual with DMD are ~10x higher than the general population.³⁷ These direct healthcare costs are only a part of the overall financial burden; indirect costs, such as loss of work and difficulty finding and funding alternative care providers, increases the financial burden on families.³⁷

In our study, over one-half of caregivers identified unmanaged psychosocial issues experienced by the diagnosed individual. These needs included issues related to transitioning from childhood to adulthood. Caregiver responses showed gaps in management of age-appropriate discussion of issues relating to sexuality and information about transitioning to independent living, advance directives, and employment for the diagnosed individual. Many of these issues were addressed in a recent European Neuromuscular Centre (ENMC) workshop that aimed to identify gaps in knowledge about an emerging population of adults with DMD;²¹ considerations for management of this transitional period is also expanded in the updated DMD care considerations publication.³⁸ Additional psychosocial issues identified by caregivers in our study included counseling for the diagnosed individual, which was identified as a need by nearly three-quarters of caregivers, overall, but only one-half reported this was managed. Internalizing (e.g. depression and anxiety) and externalizing (e.g. aggression, hyperactivity) neurobehavioral problems have been highlighted in several recent publications^{39–41} and have been identified for older individuals with DMD as an area of importance that needs further attention.²¹ The failure to meet adequately the mental health needs of these individuals might be due to a lack of recognition and referral on the part of

medical professionals, but might also reflect the national need for mental health service providers.⁴²

Caregivers in our study also responded to having unmanaged needs for their own support. Family support groups and personal counseling were identified as needs by over two-thirds of caregivers, but less than one-half adequately managed these needs. The lack of adequate supports, both financial and emotional, may contribute to greater perceived burden and psychological distress for caregivers of individuals with a dystrophinopathy.^{23,26,30} A survey completed by 1238 women in the United States who were caring for someone with DMD reported an age-adjusted rate of serious psychological stress of 12.4% among caregivers, significantly higher than the 7.1% in the general population of mothers;²³ almost one-half of these women reported high levels of caregiving demands. More advanced disease is often associated with increased caregiver stress.^{26,27,43,44} Consistent with this observation, counseling for caregivers was identified as a need for a significantly higher proportion of caregivers when the diagnosed individual was non-ambulatory. Social support, resiliency, increased perceived control, and healthy family identity are factors that have been associated with less caregiver stress.^{23,27,45} Our study, along with others, suggests that recommendations for the management of psychosocial issues have not been effectively translated at point of care,^{46,47} despite evidence that adequate resources have been shown to protect against related stresses.^{27,45}

Our study is not without limitations. The perceived needs and their management were from the perspective of the caregiver. Individuals with dystrophinopathy are increasingly voicing their desire to have their stories told from their own perspective as they approach adulthood while living with a chronic health condition and physical disability.^{48,49} To collect these perspectives, there is a need for both young-person-friendly and caregiver-free data collection methods to ensure an unbiased assessment. A recent article from MD STARnet utilized data from an Internet-based questionnaire that collected the perspectives of life transitions and health services among individuals aged 16 years and older.⁵⁰ The response rate was low (25%), but of those who responded,

gaps in services were consistent with those reported by the caregiver in this study. Another limitation is the geographical restriction of the MD STARnet sites and, as noted previously, caregivers responding to our survey were more likely to be non-Hispanic Whites and have higher education levels than all eligible MD STARnet caregivers combined. Thus, our survey findings might not represent the needs of all families identified by the MD STARnet or the wider population of families affected by a dystrophinopathy diagnosis across the United States. Further, surveys were collected over an extended period. Nonetheless, our findings are consistent with recent related publications suggesting current relevance. A final limitation is the inclusion of caregivers of DMD and BMD, which vary in clinical severity, and thus, may result in different disease management needs. However, our stratification by current ambulation status, which could be a more salient indicator of need, probably captured the association with disease severity that might emerge if the stratification was done by phenotype. Despite these limitations, our study has advantages over available clinic-based studies, including greater representativeness of all families affected by a dystrophinopathy diagnosis. Furthermore, our study included a broad range of needs across multiple domains and the lifespan allowing a more comprehensive assessment of areas needing additional resources and guidance from the clinical community.

In summary, the findings from our study are of value to providers, payers, and policymakers as they work together to meet the many needs identified by caregivers of individuals with dystrophinopathies. It is reassuring that families indicate their needs around medical care and disease-related information are generally being met. Additional resources and policies might focus on the unmet financial and psychosocial needs of this population, as recognized in the recently updated care considerations.³⁸

Conclusion

Caregivers of patients diagnosed with a dystrophinopathy report diverse needs for the patient and the family in the management of the disease. Our study showed that needs associated with physical health and access to information were managed by most families. Less managed areas identified by our survey included funding for needs not covered by insurance and psychosocial support. We also demonstrated variability in needs and their management by ambulation status of the patient, household income, and caregiver race/ethnicity. Continued advocacy on behalf of patients with a dystrophinopathy to healthcare providers, public health practitioners, and policymakers should focus on addressing gaps in provision of needed financial assistance and psychosocial services.

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Declaration of conflicting interests

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Ethical approval

Each site received institutional review board (IRB) approval for the caregiver survey (University of Arizona IRB-1, protocol 05-0426-01; Colorado Department of Public Health and Environment IRB, protocol 2006001; Georgia Department of Public Health IRB, protocol 090805; Centers for Disease Control and Prevention IRB-A, protocol 4792; University of Iowa IRB-1, protocol 200509724; New York State Department of Health, protocol 03-062).

Ethical publication statement

We confirm that we have read the journal's position on issues involved in ethical publication and affirm that this report is consistent with those guidelines.

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Informed consent


The University of Arizona IRB-1, Georgia Department of Public Health IRB and Centers for Disease Control and Prevention IRB-A, University of Iowa IRB-1, and New York State Department of Health waived the requirement of signed consent and approved implied consent if the caregiver completed and returned the survey. Written consent was required by the Colorado Department of Public Health and Environment IRB for Colorado caregivers.

Supplemental material

Supplemental material for this article is available online.

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References

- Emery AE. The muscular dystrophies. *Lancet* 2002; 359(9307): 687–695.
- Balaban B, Matthews DJ, Clayton GH, et al. Corticosteroid treatment and functional improvement in Duchenne muscular dystrophy: long-term effect. *Am J Phys Med Rehabil* 2005; 84(11): 843–850.
- Barber BJ, Andrews JG, Lu Z, et al. Oral corticosteroids and onset of cardiomyopathy in Duchenne muscular dystrophy. *J Pediatr* 2013; 163(4): 1080–1084.e1.
- Biggar WD, Harris VA, Eliasoph L, et al. Long-term benefits of deflazacort treatment for boys with Duchenne muscular dystrophy in their second decade. *Neuromuscul Disord* 2006; 16(4): 249–255.
- Houde S, Filiatrault M, Fournier A, et al. Deflazacort use in Duchenne muscular dystrophy: an 8-year follow-up. *Pediatr Neurol* 2008; 38(3): 200–206.
- King WM, Ruttencutter R, Nagaraja HN, et al. Orthopedic outcomes of long-term daily corticosteroid treatment in Duchenne muscular dystrophy. *Neurology* 2007; 68(19): 1607–1613.
- Lebel DE, Corston JA, McAdam LC, et al. Glucocorticoid treatment for the prevention of scoliosis in children with Duchenne muscular dystrophy: long-term follow-up. *J Bone Joint Surg Am* 2013; 95(12): 1057–1061.
- Manzur AY, Kuntzer T, Pike M, et al. Glucocorticoid corticosteroids for Duchenne muscular dystrophy. *Cochrane Database Syst Rev* 2008; 1: CD003725.
- Markham LW, Kinnett K, Wong BL, et al. Corticosteroid treatment retards development of ventricular dysfunction in Duchenne muscular dystrophy. *Neuromuscul Disord* 2008; 18(5): 365–370.
- Markham LW, Spicer RL, Khoury PR, et al. Steroid therapy and cardiac function in Duchenne muscular dystrophy. *Pediatr Cardiol* 2005; 26(6): 768–771.
- Matthews E, Brassington R, Kuntzer T, et al. Corticosteroids for the treatment of Duchenne muscular dystrophy. *Cochrane Database Syst Rev* 2016; 5: CD003725.
- Moxley RT 3rd, Pandya S, Cifaloni E, et al. Change in natural history of Duchenne muscular dystrophy with long-term corticosteroid treatment: implications for management. *J Child Neurol* 2010; 25(9): 1116–1129.
- Silversides CK, Webb GD, Harris VA, et al. Effects of deflazacort on left ventricular function in patients with Duchenne muscular dystrophy. *Am J Cardiol* 2003; 91(6): 769–772.
- Moxley RT 3rd, Ashwal S, Pandya S, et al. Practice parameter: corticosteroid treatment of Duchenne dystrophy: report of the Quality Standards Subcommittee of the American Academy of Neurology and the Practice Committee of the Child Neurology Society. *Neurology* 2005; 64(1): 13–20.
- Henricson EK, Abresch RT, Cnaan A, et al. The cooperative international neuromuscular research group Duchenne natural history study: glucocorticoid treatment preserves clinically meaningful functional milestones and reduces rate of disease progression as measured by manual muscle testing and other commonly used clinical trial outcome measures. *Muscle Nerve* 2013; 48(1): 55–67.
- Gloss D, Moxley RT 3rd, Ashwal S, et al. Practice guideline update summary: corticosteroid treatment of Duchenne muscular dystrophy: report of the Guideline Development Subcommittee of the American Academy of Neurology. *Neurology* 2016; 86(5): 465–472.
- Birnkrant DJ, Bushby K, Bann CM, et al. Diagnosis and management of Duchenne muscular dystrophy, part 2: respiratory, cardiac, bone health, and orthopaedic management. *Lancet Neurol* 2018; 17(4): 347–361.
- Birnkrant DJ, Bushby K, Bann CM, et al. Diagnosis and management of Duchenne muscular dystrophy, part 1: diagnosis, and neuromuscular, rehabilitation, endocrine, and gastrointestinal and nutritional management. *Lancet Neurol* 2018; 17(3): 251–267.
- Schreiber-Katz O, Klug C, Thiele S, et al. Comparative cost of illness analysis and assessment of health care burden of Duchenne and Becker muscular dystrophies in Germany. *Orphanet J Rare Dis* 2014; 9: 210.
- Ryder S, Leadley RM, Armstrong N, et al. The burden, epidemiology, costs and treatment for Duchenne muscular dystrophy: an evidence review. *Orphanet J Rare Dis* 2017; 12(1): 79.
- Rahbek J, Steffensen BF, Bushby K, et al. 206th ENMC International Workshop: care for a novel group of patients—adults with Duchenne muscular dystrophy Naarden, The Netherlands, 23-25 May 2014. *Neuromuscul Disord* 2015; 25(9): 727–738.
- Anderson M, Elliott EJ and Zuryski YA. Australian families living with rare disease: experiences of diagnosis, health services use and needs for psychosocial support. *Orphanet J Rare Dis* 2013; 8: 22.
- Kenneson A and Bobo JK. The effect of caregiving on women in families with Duchenne/Becker muscular dystrophy. *Health Soc Care Community* 2010; 18(5): 520–528.
- Ouyang L, Grosse SD and Kenneson A. Health care utilization and expenditures for children and young adults with muscular dystrophy in a privately insured population. *J Child Neurol* 2008; 23(8): 883–888.
- Nereo NE, Fee RJ and Hinton VJ. Parental stress in mothers of boys with Duchenne muscular dystrophy. *J Pediatr Psychol* 2003; 28(7): 473–484.
- Pangalila RF, van den Bos GA, Stam HJ, et al. Subjective caregiver burden of parents of adults with Duchenne muscular dystrophy. *Disabil Rehabil* 2012; 34(12): 988–996.
- Frishman N, Conway KC, Andrews J, et al. Perceived quality of life among caregivers of children with a childhood-onset dystrophinopathy: a double ABCX model of caregiver stressors and perceived resources. *Health Qual Life Outcomes* 2017; 15(1): 33.
- Landfeldt E, Lindgren P, Bell CF, et al. The burden of Duchenne muscular dystrophy: an international, cross-sectional study. *Neurology* 2014; 83(6): 529–536.

29. Larkindale J, Yang W, Hogan PF, et al. Cost of illness for neuromuscular diseases in the United States. *Muscle Nerve* 2014; 49(3): 431–438.
30. Magliano L, D'Angelo MG, Vita G, et al. Psychological and practical difficulties among parents and healthy siblings of children with Duchenne vs. Becker muscular dystrophy: an Italian comparative study. *Acta Myol* 2014; 33(3): 136–143.
31. De Moura MC, Wutzki HC, Voos MC, et al. Is functional dependence of Duchenne muscular dystrophy patients determinant of the quality of life and burden of their caregivers. *Arg Neuropsiquiatr* 2015; 73(1): 52–57.
32. Chen JY and Clark MJ. Family function in families of children with Duchenne muscular dystrophy. *Fam Community Health* 2007; 30(4): 296–304.
33. Chen JY and Clark MJ. Family resources and parental health in families of children with Duchenne muscular dystrophy. *J Nurs Res* 2010; 18(4): 239–248.
34. Miller LA, Romitti PA, Cunniff C, et al. The Muscular Dystrophy Surveillance Tracking and Research Network (MD STARnet): surveillance methodology. *Birth Defects Res A Clin Mol Teratol* 2006; 76(11): 793–797.
35. Mathews KD, Cunniff C, Kantamneni JR, et al. Muscular Dystrophy Surveillance Tracking and Research Network (MD STARnet): case definition in surveillance for childhood-onset Duchenne/Becker muscular dystrophy. *J Child Neurol* 2010; 25(9): 1098–1102.
36. The American Association for Public Opinion Research (AAPOR). Standard definitions: final disposition of case codes and outcome rates for surveys. 7th ed., [https://www.aapor.org/Standards-Ethics/Standard-Definitions-\(1\).aspx](https://www.aapor.org/Standards-Ethics/Standard-Definitions-(1).aspx) (2011, accessed 20 November 2011).
37. Cavazza M, Kodra Y, Armeni P, et al. Social/economic costs and health-related quality of life in patients with Duchenne muscular dystrophy in Europe. *Eur J Health Econ* 2016; 17(Suppl. 1): 19–29.
38. Birnkrant DJ, Bushby K, Bann CM, et al. Diagnosis and management of Duchenne muscular dystrophy, part 3: primary care, emergency management, psychosocial care, and transitions of care across the lifespan. *Lancet Neurol* 2018; 17(5): 445–455.
39. Banihani R, Smile S, Yoon G, et al. Cognitive and neurobehavioral profile in boys with Duchenne muscular dystrophy. *J Child Neurol* 2015; 30(11): 1472–1482.
40. Ricotti V, Mandy WP, Scoto M, et al. Neurodevelopmental, emotional, and behavioural problems in Duchenne muscular dystrophy in relation to underlying dystrophin gene mutations. *Dev Med Child Neurol* 2016; 58(1): 77–84.
41. Caspers Conway K, Mathews KD, Paramsothy P, et al. Neurobehavioral concerns among males with dystrophinopathy using population-based surveillance data from the Muscular Dystrophy Surveillance, Tracking, and Research Network. *J Dev Behav Pediatr* 2015; 36(6): 455–463.
42. Hoge MA, Morris JA, Stuart GW, et al. A national action plan for workforce development in behavioral health. *Psychiatr Serv* 2009; 60(7): 883–887.
43. Magliano L, Patalano M, Sagliocchi A, et al. Burden, professional support, and social network in families of children and young adults with muscular dystrophies. *Muscle Nerve* 2015; 52(1): 13–21.
44. Landfeldt E, Lindgren P, Bell CF, et al. Quantifying the burden of caregiving in Duchenne muscular dystrophy. *J Neurol* 2016; 263(5): 906–915.
45. Peay HL, Meiser B, Kinnett K, et al. Mothers' psychological adaptation to Duchenne/Becker muscular dystrophy. *Eur J Hum Genet* 2016; 24(5): 633–637.
46. Bushby K, Finkel R, Birnkrant DJ, et al. Diagnosis and management of Duchenne muscular dystrophy, part 2: implementation of multidisciplinary care. *Lancet Neurol* 2010; 9(2): 177–189.
47. Bushby K, Finkel R, Birnkrant DJ, et al. Diagnosis and management of Duchenne muscular dystrophy, part 1: diagnosis, and pharmacological and psychosocial management. *Lancet Neurol* 2010; 9(1): 77–93.
48. Gibson BE, Mistry B, Smith B, et al. Becoming men: gender, disability, and transitioning to adulthood. *Health* 2014; 18(1): 95–114.
49. Hamdani Y, Mistry B and Gibson BE. Transitioning to adulthood with a progressive condition: best practice assumptions and individual experiences of young men with Duchenne muscular dystrophy. *Disabil Rehabil* 2015; 37(13): 1144–1151.
50. Paramsothy P, Herron AR, Lamb M, et al. Health care transition experiences of males with childhood-onset Duchenne and Becker muscular dystrophy: findings from the Muscular Dystrophy Surveillance Tracking and Research Network (MD STARnet) Health Care transitions and other life experiences survey. *PLoS Curr*. Epub ahead of print 21 August 2018. DOI: 10.1371/currents.md.7de8a1c6798d7a48d38ea09bd624e1cd.