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Characterization of individuals with selected muscular dystrophies from the expanded pilot of the Muscular Dystrophy Surveillance, Tracking and Research Network (MD STARnet) in the United States

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Abstract

Introduction: Data on muscular dystrophies (MDs), a heterogeneous group of heritable diseases hallmarked by progressive muscle deterioration, are scarce.

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AUTHOR CONTRIBUTIONS

Bailey Wallace led the development of this manuscript, completed the primary analysis, conceptualized and drafted the manuscript. K. Tiffany Smith contributed to concept design and development of the manuscript. Shiny Thomas completed the secondary analysis. Natalie Street supervised the manuscript development. Kristin M. Conway contributed to concept and design and manuscript revisions. Christina Westfield reviewed manuscript versions and tables, provided clinical feedback and historical background. Jennifer G. Andrews reviewed several manuscript versions and tables. Richard O. Weinert reviewed the manuscript and contributed to the acquisition of the data. Thuy Quynh N. Do contributed to study concept and design and manuscript revisions. All authors reviewed the final manuscript, provided extensive feedback on its content, and approved its submission.

CONFLICT OF INTEREST

The authors have no potential conflicts of interest.

DATA AVAILABILITY STATEMENT

Due to privacy concerns, data from MD STARnet pilot is not publicly available. We are willing to work with individuals who would like to verify the results. Data from this analysis is kept at the Centers for Disease Control and Prevention. Researchers interested in utilizing MD STARnet data may obtain a data request form and further instructions on data access and sharing at www.cdc.gov/ncbddd/musculardystrophy/accessing-md-starnet-data.html. For more information on data access, please email MDSTARnet@CDC.gov.

SUPPORTING INFORMATION

Additional supporting information may be found online in the Supporting Information section at the end of this article.

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Objective: We describe cross-sectional sociodemographic and clinical characteristics of individuals with congenital, distal, Emery-Dreifuss, facioscapulohumeral, limb-girdle, myotonic, or oculopharyngeal MD.

Methods: The study was conducted in four sites (Arizona, Colorado, Iowa, and 12 western New York counties) as a pilot expansion of the Muscular Dystrophy Surveillance, Tracking and Research Network, funded by the Centers for Disease Control and Prevention. MDs were detected in healthcare facilities and administrative data sources using International Classification of Disease codes. Our sample contains 1,723 individuals with a MD diagnosis and a healthcare encounter between January 1, 2007 and December 31, 2011.

Results and Conclusions: Individuals were mostly non-Hispanic and white. Median ages ranged from 9.2 to 66.0 years. Most (98%) had health insurance. The proportion of individuals who were disabled or unable to work increased with age (range: 8.6–46.4%). People with limb-girdle MD aged ≥18 years were more likely to be nonambulatory (range: 24.5–44.7%). The percentages of individuals with documented clinical interventions during the surveillance period were low. The most common cause of death was respiratory causes (46.3–57.1%); an ICD-10 code for MD (G71.1 or G71.0) was reported for nearly one-half. Our findings show wide variability in sociodemographic and clinical characteristics across MDs.

Keywords

epidemiology; MD STAR*net*; muscular dystrophy; population-based; surveillance

1 | INTRODUCTION

Muscular dystrophies (MDs) are a group of disorders characterized by progressive weakness and muscle wasting that vary in age of onset, severity, and genetic cause (Mercuri & Muntoni, 2013). The breadth and diversity of symptoms requires complex disease management, which has resulted in care recommendations for some, but not all, MDs (Ashizawa et al., 2018; Kang et al., 2015; Narayanaswami et al., 2014; Tawil et al., 2015). The global prevalence of MD is 16 per 100,000 (Mah et al., 2016) with regional variations in prevalence for some MD types (Carter, Sheehan, Prochoroff, & Birnkrant, 2018; Fu & Xiong, 2017; Mah et al., 2016).

Understanding sociodemographic and clinical attributes of persons affected by MD is essential for assessing health needs. Although U.S. population-based data for Duchenne (DMD) and Becker (BMD) MDs exist (Sahay et al., 2019), much of the literature for other types of MDs in the United States use data from registries, clinics, and small studies. The Muscular Dystrophy Surveillance, Tracking and Research Network (MD STAR*net*), funded by the Centers for Disease Control and Prevention (CDC), is the only multi-site population-based surveillance system for MD in the United States (Sahay et al., 2019). MD STAR*net* has conducted longitudinal surveillance for DMD and BMD since 2002 (Mathews et al., 2010; Miller et al., 2006). From 2011 to 2014, a cross-sectional pilot study was conducted to examine the feasibility of expanding MD STAR*net* surveillance to include seven additional MDs (congenital [CMD], distal [DD], Emery-Dreifuss [EDMD], facioscapulohumeral [FSHD], limb-girdle [LGMD], myotonic [DM], and oculopharyngeal [OPMD]) (Do et al.,

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2018). This paper describes sociodemographic and clinical characteristics of individuals with these seven MD types in four U.S. sites using data from the MD STAR*net* pilot study.

2 | METHODS

We conducted a cross-sectional analysis of population-based data collected retrospectively from the medical records of individuals with MD. Details of the surveillance methods used by the MD STAR*net* pilot have previously been described (Do et al., 2018). Briefly, surveillance activities were conducted in four sites: Arizona (AZ), Colorado (CO), Iowa (IA), and 12 counties in western New York (wNY). Individuals were identified in administrative and healthcare sources using International Classification of Diseases (ICD) codes for congenital hereditary MD (ICD-9-CM 359.0; ICD-10 G71.0), hereditary progressive MD (ICD-9-CM 359.1, ICD-10 G71.0), and myotonic dystrophy (ICD-9-CM 359.21, ICD-10 G71.1). Sources of healthcare data included neuromuscular clinics, physical medicine and rehabilitation clinics, other outpatient clinics, and hospitals. Access to administrative data sources varied across sites. Administrative data sources included birth defects surveillance programs, healthcare administrative data with accounting records, hospital discharge summaries, Medicaid claims, state vital records, and the national death index (Do et al., 2018).

Individuals were considered eligible for inclusion in MD STAR*net* if they had an eligible MD diagnosis, residency in an MD STAR*net* site, and at least one healthcare encounter from January 1, 2007 through December 31, 2011. Data were extracted from medical records by trained abstractors. A committee comprised experienced neuromuscular clinicians reviewed the abstracted data to determine MD type and apply the MD STAR*net* diagnostic method, which was based on a clinical diagnosis or availability of confirmatory genetic test results in an individual or a family member. A clinical diagnosis required only a mention of the individual having a MD diagnosis in the medical record and not a pathogenic confirmation of genetic mutation in the individual or family member. Other information confirming the diagnosis could include muscle biopsy or signs and symptoms.

2.1 | Surveillance authority/IRB

The CO, IA, and wNY MD STAR*net* sites operated through public health authority for surveillance via their state health departments and received IRB approval or exemption. The AZ MD STAR*net* site operated through public health authority for surveillance via the state health department and received IRB approval or exemption at each health care facility that served as a data source.

2.2 | Inclusion criteria

We included individuals diagnosed with CMD, DD, DM, EDMD, FSHD, LGMD, or OPMD. Two individuals with DM were excluded due to contradictory information about the date of the birth in their medical records. There were no exclusions based on age or other criteria. Our cohort was comprised of: 941 individuals with DM, 278 with FSHD, 260 with LGMD, 119 with OPMD, 86 individuals with CMD, 17 with DD, and 22 with EDMD (N = 1,723).

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2.3 | Sociodemographic and clinical characteristics

The following characteristics were available for analysis: biological sex (male, female), race (white or Caucasian, black or African American, Asian, American Indian or Alaska Native, Native Hawaiian or Pacific Islander, multiple, other, unknown), ethnicity (Hispanic, not Hispanic, unknown), vital status (presumed living, living verified, deceased), method of diagnosis (clinical diagnosis, genetic diagnosis in self, or genetic diagnosis in family), age at death if deceased, reported cause of death, age at the beginning of the surveillance period if alive on January 1, 2007, insurance status (private, public, both, uninsured/self-pay, not documented), employment (working for pay, homemaker, student, retired, younger than school age, disabled or unable to work, unemployed, other, not documented), ambulatory status (able to ambulate, part-time wheelchair or scooter, part-time unknown device, full-time manual wheelchair, full-time power wheelchair, full-time unknown device, other, not documented), and the following clinical interventions (yes, not documented): percutaneous endoscopic gastrostomy (PEG) noninvasive positive pressure ventilation (NIPPV), tracheostomy, cough assist, pacemaker, defibrillator, and cardiac transplant. Racial groups with small number of individuals (American Indian/Alaska Native, Asian, multiple races, Native Hawaiian/Pacific Islander, other) were coded as Other/Multiple race.

Data on the cause of death were sourced from state death certificates, medical records, and a national death index search. The use of clinical devices or procedures was based on any use during the surveillance period but could have been initiated before this period. Time-sensitive variables, including ambulatory status, employment status, and vital status, were abstracted for the last health encounter in the surveillance period for which the information was available. The ages used to stratify ambulatory status and employment were calculated using the date at which the information was recorded in the medical record. Ambulatory status was assessed among those who were of walking age (2 years and older). The ambulatory status of individuals who were able to ambulate but used a wheel chair part-time, was coded as “ambulatory,” whereas individuals who used a device full-time, such as a manual or power wheelchair or scooter, were coded “non-ambulatory.” Employment status was assessed among those of working age, which was defined as 18–64 years of age. Individuals who were retired, homemakers, or had unknown employment were included in the Other/Not Documented employment category.

2.4 | Data analysis

We calculated means (*SD*), medians and minimum/maximum for continuous variables and frequencies and percentages for categorical variables. Demographic variables with a frequency under 5 are not reported to protect confidentiality. Analyses were conducted using SAS 9.4 © (SAS Institute, Cary, NC). FSHD, LGMD, and DM were stratified by age at last employment and ambulation status. Stratified analyses were not performed for CMD, DD, EDMD, and OPMD due to small numbers.

3 | RESULTS

Ages at the start of the study ranged from infancy to 93 years (Table 1). The percentage of deceased individuals ranged from 7.6 to 21.0%. Males and females were evenly distributed

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across MDs except for EDMD, which had a higher percentage of males. Individuals with MD were predominantly white, and the majority of individuals were non-Hispanic. However, ethnicity was unknown for nearly one-third of individuals for most MDs, except for CMD. The percentage of individuals with only public insurance ranged from 22.7% (EDMD) to 49.1% (DM). Overall, 34.9% (n = 601) of individuals had genetic confirmation of MD in self, 7.4% (n = 127) had genetic confirmation of MD through a family member, and 57.8% (n = 995) had a clinical diagnosis of MD. As previously reported, clinical diagnosis was the most common diagnostic method for the MD subtypes—except for EDMD—ranging from 49.6% for FSHD to 77.7% for LGMD (Do et al., 2018).

The median age at death ranged from middle (DM) to late adulthood (OPMD) for all MDs, except for CMD. The median age at death for the nine deceased individuals with CMD was 12.8 years (Table 1). The most common cause of death for individuals with FSHD, LGMD, or DM were respiratory causes (46.3–57.1%) (Table 2). An ICD-10 code specific to MD (G71.1 or G71.0) was reported for 62.2% of deceased individuals with DM, 57.1% of deceased individuals with FSHD, and 46.7% of deceased individuals with LGMD.

The percentage of individuals with DM who were disabled or unable to work was higher than the percentage of individuals with FSHD or LGMD at all ages (Table 3). The percentage of individuals with DM who were disabled or unable to work was highest for those aged 40–<50 years (42.9%) and 50 to 64 years (44.8%). For the MD types not stratified by age due to the small number of cases (Table S1), the lowest percentage of disabled or unable to work was found among those diagnosed with CMD (11.1%), compared to individuals with DD (13.3%), EDMD (13.3%), or OPMD (20.5%). Among individuals aged 4–18 years, enrollment in school was higher than 80% for all MDs (data not shown).

Ambulation status varied by age group and MD (Table 4). The highest percentage of non-ambulatory individuals was among those diagnosed with LGMD, especially those older than 18 years. Depending on age, up to 27.1% of individuals diagnosed with FSHD or DM were non-ambulatory. Of the remaining MDs (Table S1), individuals with CMD were more frequently non-ambulatory (43.0% vs. 17.7% for DD, 27.3% for EDMD, and 12.0% for OPMD).

Overall, the percentages of individuals with clinical interventions documented during the surveillance period were low (Table 5). NIPPV was the most broadly documented across MDs (5.0–27.9%), with the highest percentage among people with CMD. PEGs were documented for individuals with all forms of MD (1.4–25.2%), except for DD; individuals with CMD (20.9%) and OPMD (25.2%) had the highest frequency of PEG. Cardiac interventions were infrequent, with 6.6% of individuals with MD having a pacemaker, 2.4% a defibrillator and 0.5% a cardiac transplant. Of individuals with DM, 9.9% had a pacemaker and 3.8% had a defibrillator. Of individuals with EDMD, 18.2% had a pacemaker and 4.6% had a defibrillator documented in their medical record.

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4 | DISCUSSION/CONCLUSIONS

In this study, we describe key sociodemographic and clinical interventions of individuals with seven types of MD from four U.S. sites. Most individuals with MD were white and were non-Hispanic.

Overall, a clinical diagnosis with MD was the most common diagnostic method. A high percentage of individuals with DM or LGMD were disabled or unable to work. A high percentage of individuals with DM, CMD, or LGMD relied on public insurance. School enrollment did not appear to be impacted by MD (data not shown). The percentages of deceased individuals ranged from 7.6 to 21%. The reported mean ages of death for individuals with DM (53.6 years) and FSHD (66.1 years) in this study are similar to those reported by a U.S. registry study (DM Type 1 = 55.7 and FSHD = 64.0 years) (Hilbert et al., 2012). The most frequent cause of death was respiratory causes for people with FSHD, LGMD, or DM, and close to one-half of deceased individuals had an ICD-10 code for MD included in their death certificate. The proportions of individuals with documented clinical interventions during our surveillance period were relatively low.

Employed persons with chronic diseases have better health and wellbeing (Minis et al., 2010). Our data suggest a pattern of more frequent unemployment and disability in individuals with DM, FSHD, or LGMD, indicating a potential need for supportive measures to sustain employment for individuals with these types of MD. Differences in the percentages of individuals with these MDs who were disabled or unable to work may be due to variation in the age distribution of the working-age individuals. Few individuals with CMD, DD, EDMD, or OPMD were of working age, prohibiting a description of employment status stratified by age. More research may be needed to understand appropriate support and accommodations pertaining to continued employment for individuals affected by MD.

Only 2.3% of individuals in our study were uninsured, which is lower than the 16.3% uninsured for the general population, according to the 2010 census (ASPE, 2011). This may be the result of more individuals with MD qualifying for public insurance than the general public. Alternatively, the lower rates may be an underestimate due to bias introduced by medical record review, adults without insurance not seeking healthcare at the same rate as insured adults (Garfield, Licata, & Young, 2014), or reflect state-specific variation in public insurance coverage.

We report on specific cardiac, respiratory, and other clinical interventions for seven types of MD. For individuals with DM or EDMD cardiac dysfunction is a predominant manifestation that may necessitate intervention. Notably, 9.9% of individuals with DM had a pacemaker and 3.8% had a defibrillator documented in their medical record, which is consistent with other research showing that between 3 and 22% of DM patients require a pacemaker or cardioverter defibrillator (Laurent et al., 2011). The reported frequency of pacemaker implantation in EDMD patients has ranged from 37 to 70% (Boriani et al., 2003), with most requiring it before the age of 40 (average: 29 years) (Limipitikul, Ong, & Tomaselli, 2017; Steckiewicz et al., 2016). Our percentage of users was below these previous reports;

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however, the median age of individuals with EDMD at the beginning of this study was 21.2 years, indicating that one-half of the cohort had not reached the average age of implantation. The lower rate of cardiac interventions may also be due to the small number of cases identified, a limited period of data abstraction, or lack of documentation in medical records reviewed during the surveillance period. For respiratory interventions, both CMD and DM had the highest percentages of individuals with NIPPV (27.9 and 22.9%) and tracheostomy (8.1 and 6.5%). Noninvasive and invasive respiratory interventions are routine for CMD (Kang et al., 2015). Respiratory failure is a major cause of morbidity in individuals with DM (Sansone et al., 2015). Other studies have estimated that between 25 and 28% of DM patients need noninvasive ventilation (Bianchi et al., 2014; Pincherle et al., 2012).

The percentage of any clinical intervention was low for individuals with FSHD. This may be related to disease severity or incomplete capture of clinical interventions needed to manage common morbidities of FSHD (Balatsouras, Korres, Manta, Panousopoulou, & Vassilopoulos, 2007; Evangelista et al., 2016; Nikolic et al., 2016). Current MD STAR*net* data collection includes abstraction of data on scoliosis surgery, hearing aids, retinal diseases, and scapular fixation, which are expected to yield more complete data on the clinical management of FSHD. Given the burden of dysphagia in OPMD (Youssof, Romero-Clark, Warner, & Plowman, 2017), it is not unexpected that 25.2% of individuals with OPMD had PEG documented in the medical record.

There are several limitations in this pilot surveillance study. Although the MD STAR*net* conducts population-based surveillance, our results represent populations who received treatment within these four sites and are mostly non-Hispanic White. While the percentage of non-Hispanic cases and white cases is reflective of the geographic populations of the four sites, results may not be generalizable to the entire U.S. population. For the rarest MDs, small counts prohibited descriptions of sociodemographic variables. We retrospectively ascertained cases and were limited to the available information in clinical and vital records data. We were unable to access private neurology offices or referring physicians' offices identified in hospital records because of time and resource constraints. Consequently, our results may underestimate outcomes. In addition, data for cause of death were identified using multiple sources, that is, medical record, death certificates, and a National Death Index Search. Given the various data sources used for cause of death and the lack of uniformity between sources and how surveillance sites recorded death data, we could not parse out primary, contributing and underlying cause of death in our data. Individuals with MD who did not seek care in neuromuscular clinics, resided in an MD STAR*net* site but sought care out-of-state, or had milder symptoms requiring less frequent care may not be included in the MD STAR*net* surveillance cohort. Diagnostic delays are common for many types of MD (Hilbert et al., 2013; Menezes et al., 2012; Pogoryelova et al., 2018; Salort-Campana et al., 2015); thus, individuals not yet diagnosed during our abstraction period were not represented in this cohort. Assessing the accuracy of the diagnosis to determine MD subtype was not possible for those without definitive diagnostic testing.

This study describes sociodemographic and clinical characteristics of individuals affected by seven types of MDs in four U.S sites using cross-sectional, population-based, pilot data. Population-level descriptions of the U.S. MD populations are an essential precursor to public

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health studies, tertiary prevention, and management of available resources. Population-based, longitudinal data are needed to investigate prevalence, survival, disease progression, healthcare utilization, and disparities in care. Since the pilot, MD STAR*net* has expanded the number of surveillance areas and is collecting longitudinal clinical data to facilitate future studies on these topics.

Supplementary Material

Refer to Web version on PubMed Central for supplementary material.

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TABLE 1

Demographic characteristics in the medical record for individuals with muscular dystrophy, MD STAR*net* 2007–2011 (*N* = 1,723)

	CMD (*n* = 86)	Distal (*n* = 17)	EDMD (*n* = 22)	FSHD (*n* = 278)	LGMD (*n* = 260)	DM (*n* = 941)	OPMD (*n* = 119)
Age on 1/1/2007 (years)							
Alive on 1/1/2007 (*n*)	77	17	22	277	257	918	119
Mean (*SD*)	14.8 (14.4)	43.9 (12.6)	23.2 (14.7)	43.8 (19.9)	38.9 (21.5)	39.5 (17.5)	65.7 (9.8)
Median	9.2	40.9	21.2	48.7	38.8	41.3	66.0
min, max	0.3, 75.8	27.3, 70.3	0.2, 58.3	1.5, 78.7	0, 93.4	0, 87.1	42.2, 88.6
Age at death (in years)							
Deceased, *n* (%)	9 (10.5)	NR	NR	21 (7.6)	30 (11.5)	164 (17.4)	25 (21.0)
Mean (*SD*) min, max	12.2 (7.5)	NR	NR	65.6 (8.5)	66.8 (15.8)	53.9 (16.2)	77.0 (8.7)
Median	12.8	NR	NR	66.1	71.0	56.4	77.9
Min, max	0.0, 20.6	NR	NR	53.1, 81.2	22.0, 93.9	0.0, 91.7	61.8, 90.8
Sex, *n* (%)							
Female	41 (47.7)	10 (58.8)	5 (22.7)	117 (42.1)	129 (49.6)	468 (49.7)	65 (54.6)
Male	45 (52.3)	7 (41.2)	17 (77.3)	161 (57.9)	131 (50.3)	473 (50.3)	54 (45.4)
Race, *n* (%)							
White	66 (76.7)	12 (70.6)	21 (95.5)	228 (82.0)	202 (77.7)	803 (85.3)	75 (63.0)
Black	NR	NR	NR	NR	14 (5.4)	23 (2.4)	0 (0)
Other/multiple	12 (14.0)	NR	NR	NR	16 (6.2)	44 (4.7)	13 (10.9)
Unknown	NR	NR	NR	31 (11.2)	28 (10.8)	71 (7.6)	31 (26.1)
Ethnicity, *n* (%)							
Hispanic	19 (22.1)	NR	NR	22 (7.9)	30 (11.5)	78 (8.3)	41 (34.5)
Non-Hispanic	58 (67.4)	9 (52.9)	20 (90.9)	159 (57.2)	143 (55.0)	589 (62.6)	31 (26.1)
Unknown	9 (10.5)	NR	NR	97 (34.9)	87 (33.5)	274 (29.1)	47 (39.5)
Insurance status, *n* (%)							
Private	32 (37.2)	7 (41.2)	13 (59.1)	127 (45.7)	118 (45.4)	328 (34.9)	41 (34.5)
Public	36 (41.9)	5 (29.4)	5 (22.7)	92 (33.1)	102 (39.2)	462 (49.1)	45 (37.8)
Both[a]	16 (18.6)	2 (11.8)	0 (0.0)	25 (9.0)	27 (10.4)	82 (8.7)	29 (24.4)

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	CMD (*n* = 86)	Distal (*n* = 17)	EDMD (*n* = 22)	FSHD (*n* = 278)	LGMD (*n* = 260)	DM (*n* = 941)	OPMD (*n* = 119)
Uninsured/self-pay	0 (0.0)	0 (0.0)	2 (9.09)	9 (3.2)	6 (2.3)	22 (2.3)	0 (0.0)
Not documented	2 (2.3)	3 (17.7)	2 (0.09)	25 (9.0)	7 (2.7)	47 (5.0)	4 (3.6)

Abbreviations: CMD, congenital muscular dystrophy; DM, myotonic muscular dystrophy; EDMD, Emery-Dreifuss muscular dystrophy; FSHD, facioscapulohumeral muscular dystrophy; LGMD, limb-girdle muscular dystrophy; MD STAR*net*, Muscular Dystrophy Surveillance, Tracking, and Research Network; OPMD, oculopharyngeal muscular dystrophy. ±Cell sizes less than 5 are not reportable (NR).

[a]Both private and public insurance coverage.

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TABLE 2

Reported causes of death for individuals with muscular dystrophy, MD STAR*net* 2007–2011

	FSHD (*n* = 21, 7.6%)	LGMD (*n* = 30, 11.5%)	DM (*n* = 164, 17.4%)
Causes of death[a]			
n (%)			
Cancer	NR	NR	15 (9.2)
Cardiac	7 (33.3)	9 (30.0)	49 (29.9)
Respiratory	12 (57.1)	17 (56.7)	76 (46.3)
Stroke	NR	NR	8 (4.9)
Other	6 (28.6)	13 (43.3)	51 (31.1)
Muscular dystrophy code	12 (57.1)	14 (46.7)	102 (62.2)

Note: ± Data 5 or less are not reportable (NR).

Abbreviations: DM, myotonic muscular dystrophy; FSHD, facioscapulohumeral muscular dystrophy; LGMD, limb-girdle muscular dystrophy; MD STAR*net*, Muscular Dystrophy Surveillance, Tracking, and Research Network.

[a]Classification of the cause of death represent nonmutually exclusive categories.

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TABLE 3

Most recent known employment status documented in the medical record for individuals with muscular dystrophy of working age (18–64 years), MD STAR*net* 2007–2011

	FSHD	LGMD	DM
	***n* (%)**	***n* (%)**	***n* (%)**
18 to <30 years			
Working for pay	13 (43.3)	21 (52.5)	41 (36.0)
Disabled /unable to work	3 (10.0)	3 (7.5)	18 (15.8)
Unemployed	2 (6.7)	6 (15.0)	16 (14.0)
Other/not documented[a]	12 (40.0)	10 (25.0)	39 (34.2)
30 to <40 years			
Working for pay	19 (59.4)	13 (44.8)	58 (36.0)
Disabled/unable to work	5 (15.6)	9 (31.0)	56 (34.8)
Unemployed	3 (9.4)	2 (6.9)	18 (11.2)
Other/not documented[a]	5 (15.6)	5 (17.2)	29 (18.0)
40 to <50 years			
Working for pay	24 (61.5)	23 (53.5)	58 (28.6)
Disabled /unable to work	8 (20.5)	14 (32.6)	87 (42.9)
Unemployed	4 (10.3)	3 (7.0)	32 (15.8)
Other/not documented[a]	3 (7.7)	3 (7.0)	26 (12.8)
50 to 64 years			
Working for pay	28 (33.7)	17 (29.8)	50 (20.2)
Disabled /unable to work	27 (32.5)	20 (35.1)	111 (44.8)
Unemployed	10 (12.1)	10 (17.5)	25 (10.1)
Other/not documented[a]	18 (21.7)	10 (17.5)	62 (25.0)
Total	184 (100%)	169 (100%)	726 (100%)

Note: Excluding 17 individuals with DM, 1 with LGMD and 2 with FSHD who are missing employment age.

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Abbreviations: DM, myotonic muscular dystrophy; FSHD, facioscapulohumeral muscular dystrophy; LGMD, limb-girdle muscular dystrophy; MD STAR*net*, Muscular Dystrophy Surveillance, Tracking, and Research Network.

[a]Other or not documented employment includes people who were retired, homemakers, or those with unknown employment.

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TABLE 4

Most recent ambulatory status[a,b] documented in the medical record for individuals with muscular dystrophy of walking age (2 years or older), MD STAR*net* 2007–2011

	FSHD	LGMD	DM
Age categories	***n* (%)**	***n* (%)**	***n* (%)**
2 to <10 years			
Ambulatory	7 (100)	17 (100)	39 (84.8)
Non-ambulatory	0 (0)	0 (0)	6 (13.0)
Other/not-documented	0 (0)	0 (0)	1 (2.2)
10 to <18 years			
Ambulatory	24 (92.3)	25 (86.2)	56 (91.8)
Non-ambulatory	1 (3.9)	4 (13.8)	3 (4.9)
Other/not-documented	1 (3.9)	0 (0)	2 (3.3)
18 to <30 years			
Ambulatory	24 (82.8)	30 (71.4)	98 (87.5)
Non-ambulatory	3 (10.3)	12 (28.6)	5 (4.5)
Other/not-documented	2 (6.9)	0 (0)	9 (8.0)
30 to <45 years			
Ambulatory	44 (92.3)	37 (75.5)	224 (89.2)
Non-ambulatory	3 (6.4)	12 (24.5)	17 (6.8)
Other/not-documented	0 (0)	0 (0)	10 (4.0)
45 to <60 years			
Ambulatory	58 (68.2)	36 (53.7)	257 (86.0)
Non-ambulatory	23 (27.1)	28 (41.8)	34 (11.4)
Other/not-documented	4 (4.7)	3 (4.5)	8 (2.7)
60 to <75 years			
Ambulatory	53 (79.1)	20 (52.6)	103 (81.1)
Non-ambulatory	13 (19.4)	17 (44.7)	20 (15.8)
Other/not-documented	1 (1.5)	1 (2.6)	4 (3.2)

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Age categories	FSHD *n* (%)	LGMD *n* (%)	DM *n* (%)
75+			
Ambulatory	14 (87.5)	9 (52.9)	14 (70.0)
Non-ambulatory	1 (6.3)	6 (35.3)	5 (25.0)
Other/not-documented	1 (6.3)	2 (11.8)	1 (5.0)
Total	277 (100%)	259 (100%)	916 (100%)

Abbreviations: DM, myotonic muscular dystrophy; FSHD, facioscapulohumeral muscular dystrophy; LGMD, limb-girdle muscular dystrophy; MD STAR*net*, Muscular Dystrophy Surveillance, Tracking, and Research Network.

[a]Cases missing the age at which ambulation status was recorded in the medical record were excluded.

[b]Cases are ambulatory if they can walk with or without device support for any amount of time. Non-ambulatory cases include those who use a wheelchair full-time or who are bedridden.

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TABLE 5

Clinical Interventions documented in the medical record for individuals with muscular dystrophy, MD STAR*net* 2007–2011 (*N* = 1,723)

	Muscular dystrophy type							
Clinical interventions	**CMD (*n* = 86)**	**DD (*n* = 17)**	**EDMD (*n* = 22)**	**FSHD (*n* = 278)**	**LGMD (*n* = 260)**	**DM (*n* = 941)**	**OPMD (*n* = 119)**	**Total (*N* = 1723)**
Cough assist, *n* (%)	8 (9.3)	0 (0.0)	0 (0.0)	2 (0.7)	6 (2.3)	5 (0.5)	0 (0.0)	21 (1.2)
NIPPV[a], *n* (%)	24 (27.9)	2 (11.8)	2 (9.1)	25 (9.0)	45 (17.3)	215 (22.9)	6 (5.0)	319 (18.5)
Tracheostomy, *n* (%)	7 (8.1)	0 (0.0)	1 (4.6)	2 (0.7)	11 (4.2)	61 (6.5)	4 (3.4)	86 (5.0)
Pacemaker, *n* (%)	1 (1.2)	0 (0.0)	4 (18.2)	4 (1.4)	10 (3.9)	93 (9.9)	2 (1.7)	114 (6.6)
Defibrillator, *n* (%)	0 (0.0)	0 (0.0)	1 (4.6)	1 (0.4)	3 (1.2)	36 (3.8)	0 (0.0)	41 (2.4)
Cardiac transplant, *n* (%)	0 (0.0)	2 (11.8)	2 (9.1)	0 (0.0)	4 (1.5)	1 (0.1)	0 (0.0)	9 (0.5)
PEG, *n* (%)	18 (20.9)	0 (0.0)	1 (4.6)	4 (1.4)	10 (3.9)	101 (10.7)	30 (25.2)	164 (9.5)

Abbreviations: CMD, congenital muscular dystrophy; DD, distal muscular dystrophy; EDMD, Emery-Dreifuss muscular dystrophy; FSHD, facioscapulohumeral muscular dystrophy; DM, myotonic muscular dystrophy; LGMD, limb-girdle muscular dystrophy; MD STARnet, Muscular Dystrophy Surveillance, Tracking, and Research Network; NIPPV, noninvasive positive pressure ventilation; OPMD, oculopharyngeal muscular dystrophy; PEG, percutaneous endoscopic gastrostomy.

[a](CPAP/BIPAP).