Muscular Dystrophy Surveillance Tracking and Research Network (MD STAR*net*)

NEW YORK STATE | Department of Health

Center for Environmental Health

Bureau of Environmental and Occupational Epidemiology

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MD STAR*net*

Goals

This report summarizes results of research about Myotonic Muscular Dystrophy, Facioscapulohumeral Muscular Dystrophy, and Duchenne Muscular Dystrophy, which is based on data from the Muscular Dystrophy Surveillance, Tracking and Research Network study (MD STAR*net*). This information is relevant to clinicians, researchers, people living with muscular dystrophy, and their families and caregivers.

If you are a person living with muscular dystrophy, a family member, or caregiver, you may want to review this report with your health care provider. If you have any questions, please contact **bdls@health.ny.gov**.

Objectives

1. Share information regarding the latest developments in Myotonic Muscular Dystrophy, Facioscapulohumeral Muscular Dystrophy, and Duchenne Muscular Dystrophy with physicians, Muscular Dystrophy interest groups, people living with muscular dystrophy, and parents/caregivers of people affected by these conditions.

2. Maintain existing and develop new partnerships with stakeholders who have interests in the improvement of healthcare, healthcare access, and quality of life for people who are living with neuromuscular disorders.

MD STAR*net*

MD STAR*net* is a multi-site, population-based, surveillance system created by the Centers for Disease Control and Prevention (CDC) after the Muscular Dystrophy Community Assistance, Research and Education Act was passed in 2001.

Surveillance involves collecting data on individuals with muscular dystrophies including Myotonic Muscular Dystrophy, Facioscapulohumeral Muscular Dystrophy, Duchenne Muscular Dystrophy, and Becker Muscular Dystrophy, living or deceased, within specified geographic areas of the United States and includes longitudinal follow-up to obtain health information for these individuals. Currently, data are collected in Iowa, Virginia, South Carolina, Utah, and specific counties in Florida, North Carolina, and New York. The counties in New York include Allegany, Cattaraugus, Cayuga, Chautauqua, Chemung, Erie, Genesee, Livingston, Monroe, Niagara, Onondaga, Ontario, Orleans, Schuyler, Seneca, Steuben, Tompkins, Wayne, Wyoming, and Yates. Medical records are the primary source of data (mostly from neuromuscular clinics, but also from hospitals, neurology practices, and specialty clinics). Information is also collected from administrative sources such as Vital Records (birth and death certificates), hospital discharge data, Medicaid, and the Birth Defects Registry.

Myotonic Muscular Dystrophy

The following data are based on a cohort of 319 individuals who were diagnosed with Myotonic Muscular Dystrophy in Western NY and who had at least one healthcare visit between 1/1/2008 and 12/21/2019. This time period begins when complete data are available for Myotonic Muscular Dystrophy and goes through the end of the MD STARnet grant cycle. The data include sociodemographic characteristics such as sex, race/ethnicity, vital status, health insurance coverage, care needs, employment status, and educational attainment of individuals; and clinical characteristics such as distribution of Myotonic Muscular Dystrophy type, proportion of individuals who have at least one parent diagnosed with Myotonic Muscular Dystrophy, mobility status, signs and symptoms, and comorbidities among individuals with each type of Myotonic Muscular Dystrophy.
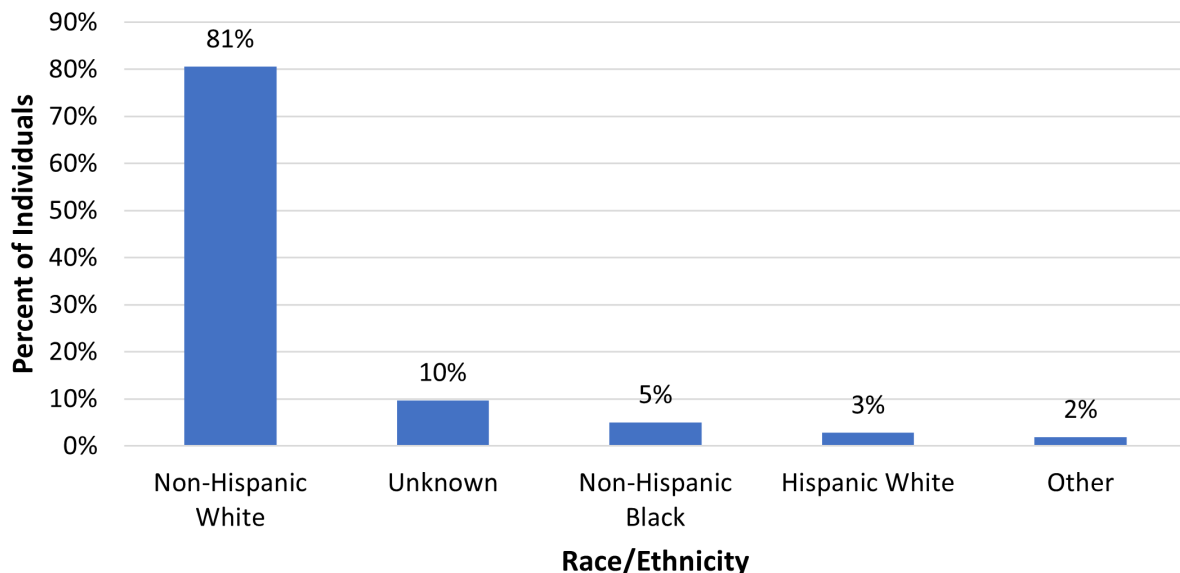
Sociodemographic Characteristics

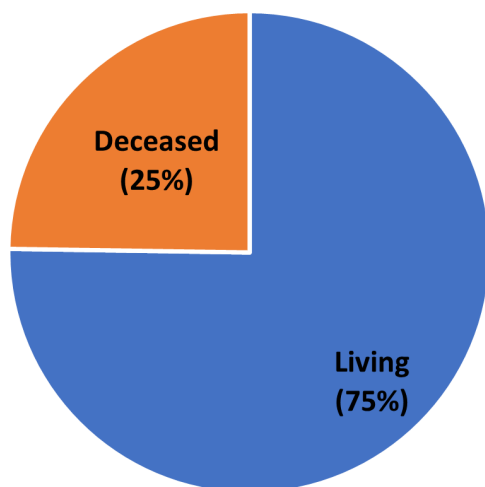**Figure 1. Sex of Individuals Diagnosed with Myotonic Muscular Dystrophy**
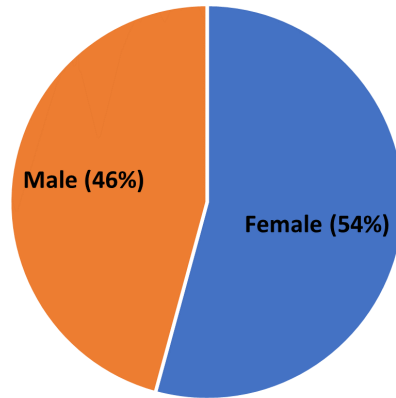
More than half of the individuals in the cohort diagnosed with Myotonic Muscular Dystrophy were females (54%).

Figure 2. Race/Ethnicity of Individuals Diagnosed with Myotonic Muscular Dystrophy


The majority of the individuals were Non-Hispanic White (81%). Race/ethnicity information was not available for 10% of the cohort.

**Figure 3. Vital Status of Individuals Enrolled in the Study with Myotonic Muscular Dystrophy**

The majority of individuals in the cohort were alive (75%) at the time of data collection.

Figure 4. Health Insurance Coverage for Individuals Diagnosed with Myotonic Muscular Dystrophy
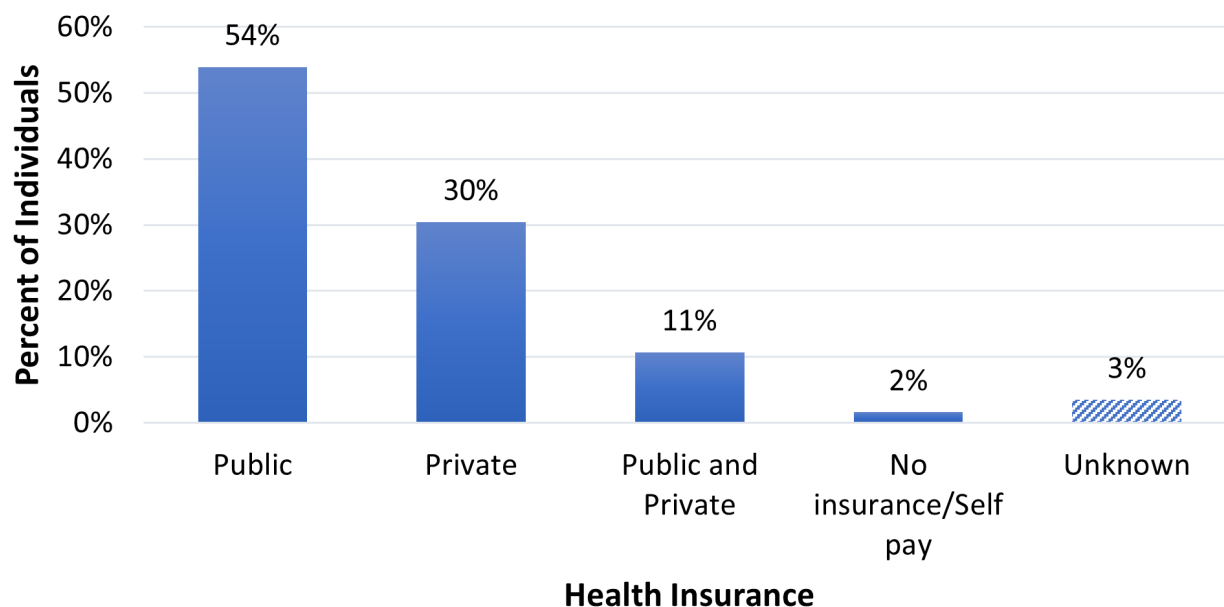

More than half of the individuals (54%) reported having public health insurance (Medicare and/or Medicaid), followed by private insurance (30%), and those who had both public and private insurance (11%). A smaller percentage (2%) of individuals had no insurance.

Figure 5. Care Needs of Individuals Diagnosed with Myotonic Muscular Dystrophy
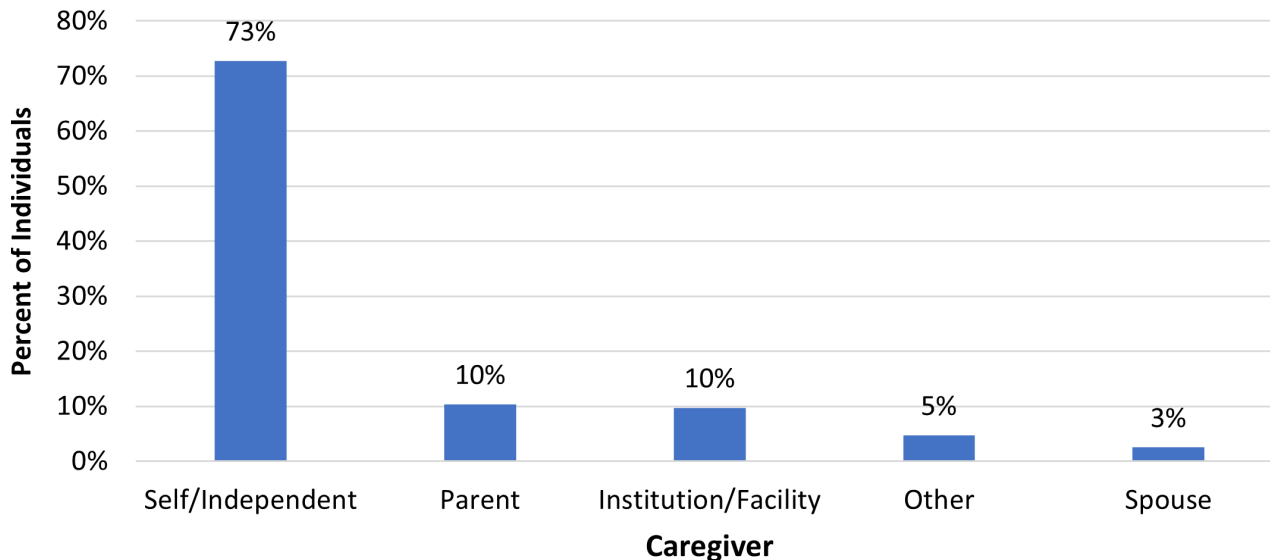
Most individuals reported caring for themselves (73%), followed by those who received care assistance from parents (10%), and those who are cared for in an institution/facility (10%).

Figure 6. Employment Status of Individuals Diagnosed with Myotonic Muscular Dystrophy
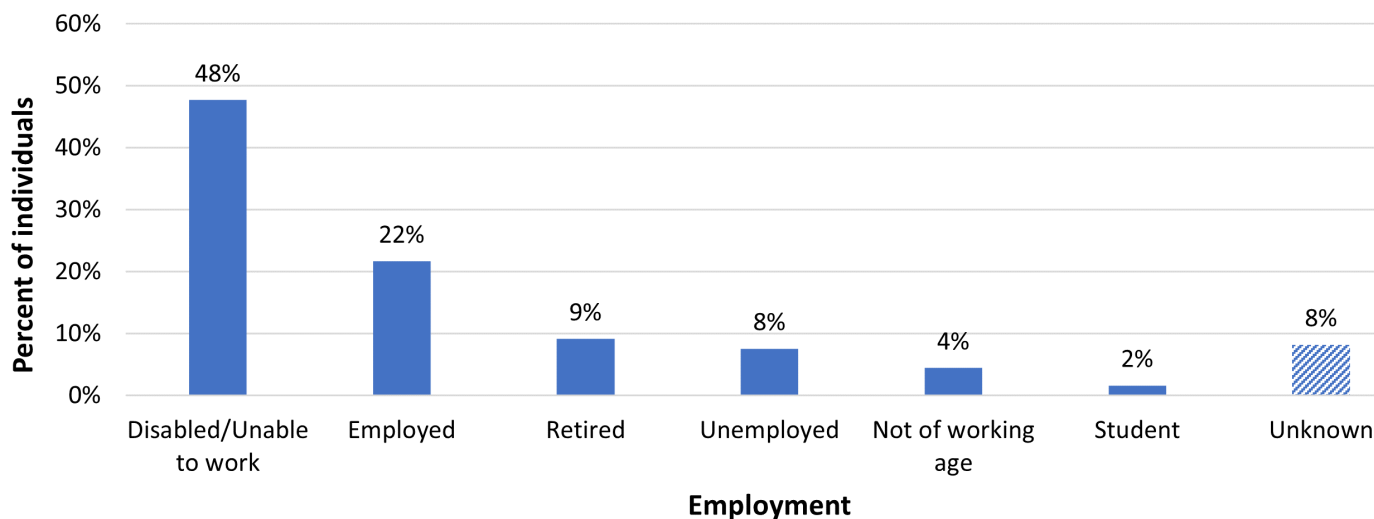
Most individuals reported being disabled and unable to work (48%), followed by those who were employed (22%) and those who were retired (9%). A smaller percentage reported being unemployed (8%), not of working age (4%), or a student (2%).

Clinical Characteristics

Table 1. Clinical Characteristics by Myotonic Muscular Dystrophy Type

Clinical Characteristics	Myotonic Muscular Dystrophy Type		
	Type 1 (min-max)	Type 2 (min-max)	Not Otherwise Specified (min-max)
Mean age at last clinic visit (in years)	44 (0-85)	63 (23-84)	48 (18-70)
Mean age ambulation ceased (in years)	50 (6-77)	69 (65-74)	53 (25-67)
Mean age at death (in years)	51 (0-86)	70 (53-81)	52 (25-66)

Figure 7. Distribution of Myotonic Muscular Dystrophy Type Among Individuals Diagnosed with Myotonic Muscular Dystrophy

Most of the individuals diagnosed with Myotonic Muscular Dystrophy had Myotonic Muscular Dystrophy Type 1 (71%).

Not Otherwise Specified* (10%)
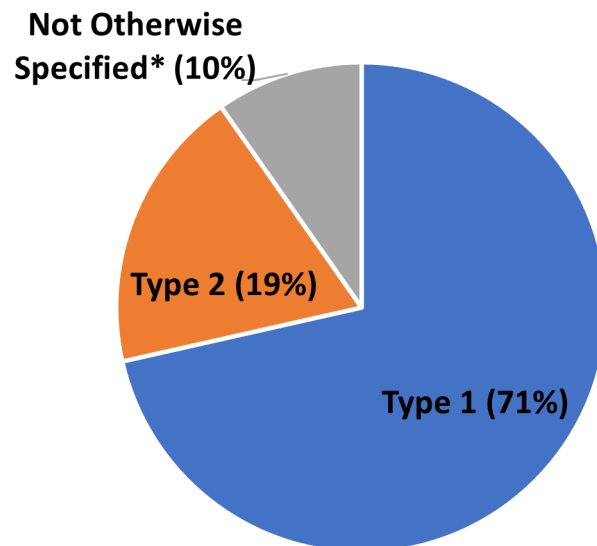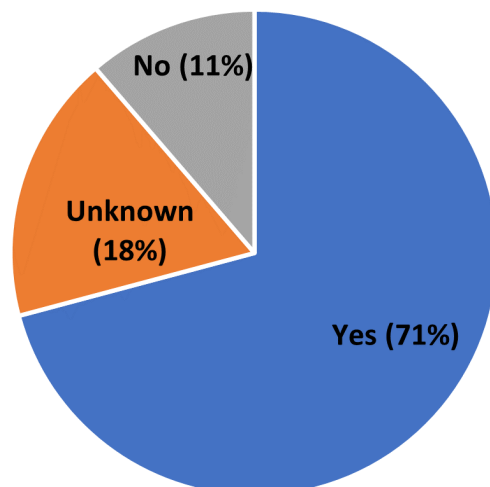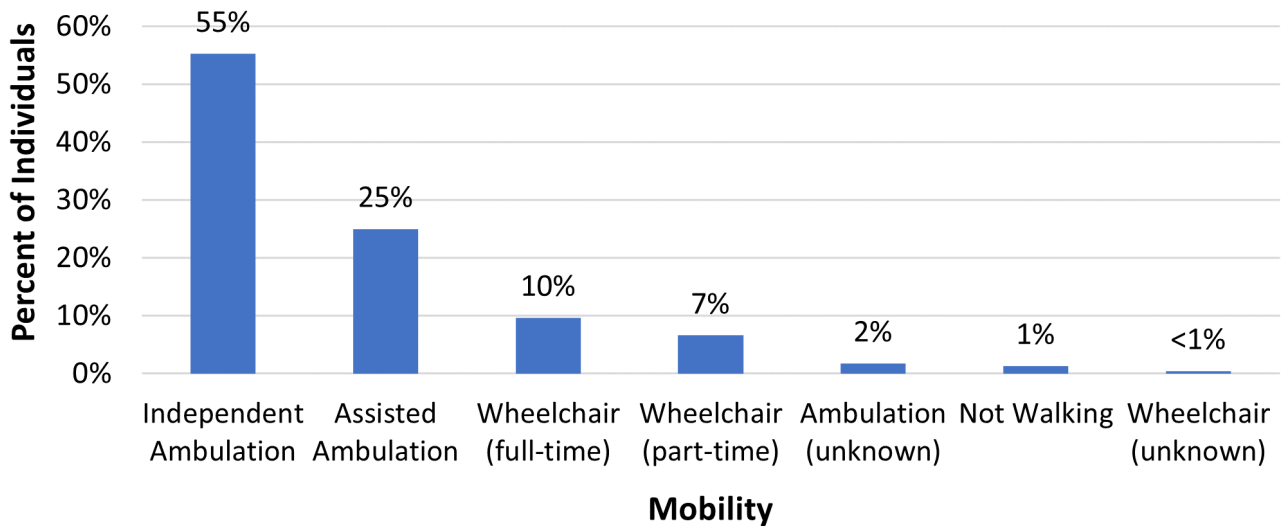
Type 2 (19%)

Type 1 (71%)

Figure 8. Proportion of Individuals Diagnosed with Myotonic Muscular Dystrophy Who Had At Least One Parent Diagnosed with Myotonic Muscular Dystrophy

Most of the individuals diagnosed with Myotonic Muscular Dystrophy had at least one parent who also had a Myotonic Muscular Dystrophy diagnosis. Information about parent diagnosis was not available for 18% of the cohort.
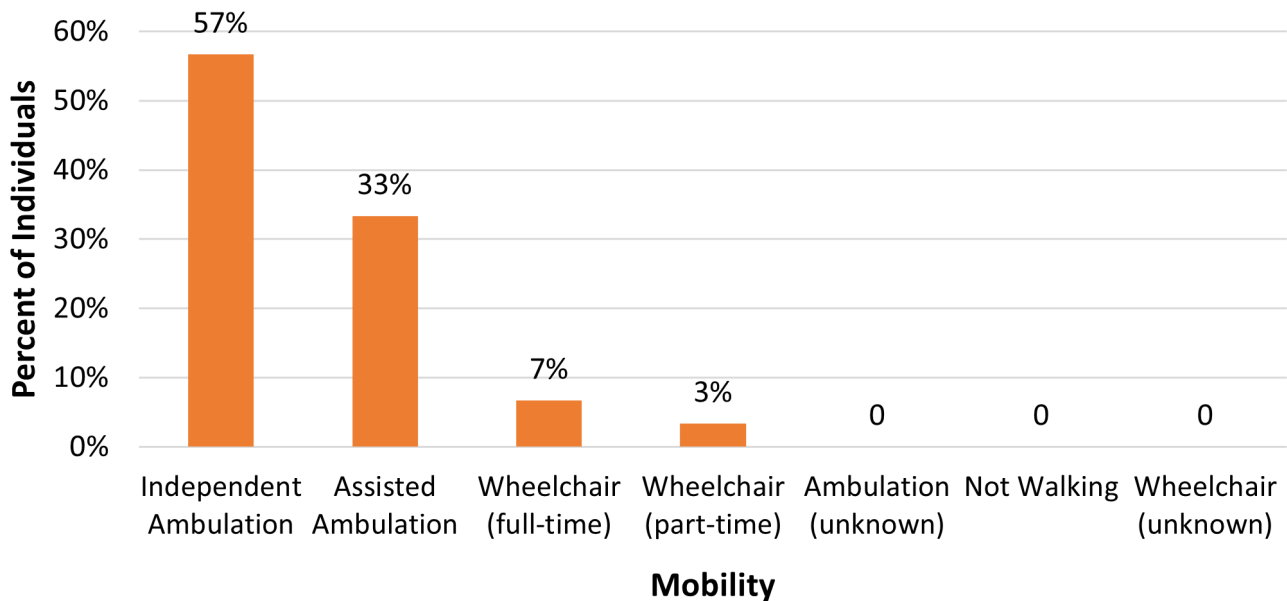
No (11%)

Unknown (18%)

Yes (71%)

Figure 9a. Mobility Status of Individuals Diagnosed with Myotonic Muscular Dystrophy Type 1

There were 228 individuals diagnosed with Myotonic Muscular Dystrophy Type 1 as of their last clinic visit. More than half (55%) reported independent ambulation. A quarter reported assisted ambulation (25%), followed by those who were wheelchair-dependent full-time (10%), and those who used a wheelchair part-time (7%). A small proportion of the individuals (1%) were infants who had not yet started to walk.
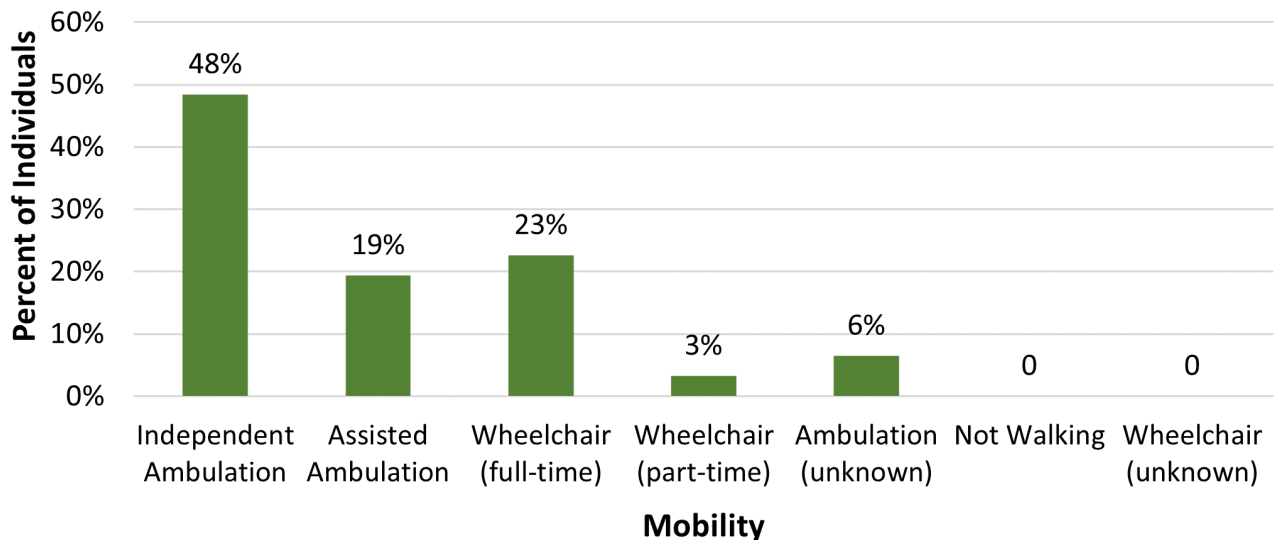
Figure 9b. Mobility Status of Individuals Diagnosed with Myotonic Muscular Dystrophy Type 2

There were 60 individuals diagnosed with Myotonic Muscular Dystrophy Type 2 as of their last clinic visit. More than half (57%) reported independent ambulation, followed by those who reported assisted ambulation (33%), those who were wheelchair-dependent full-time (7%), and those who used a wheelchair part-time (3%).
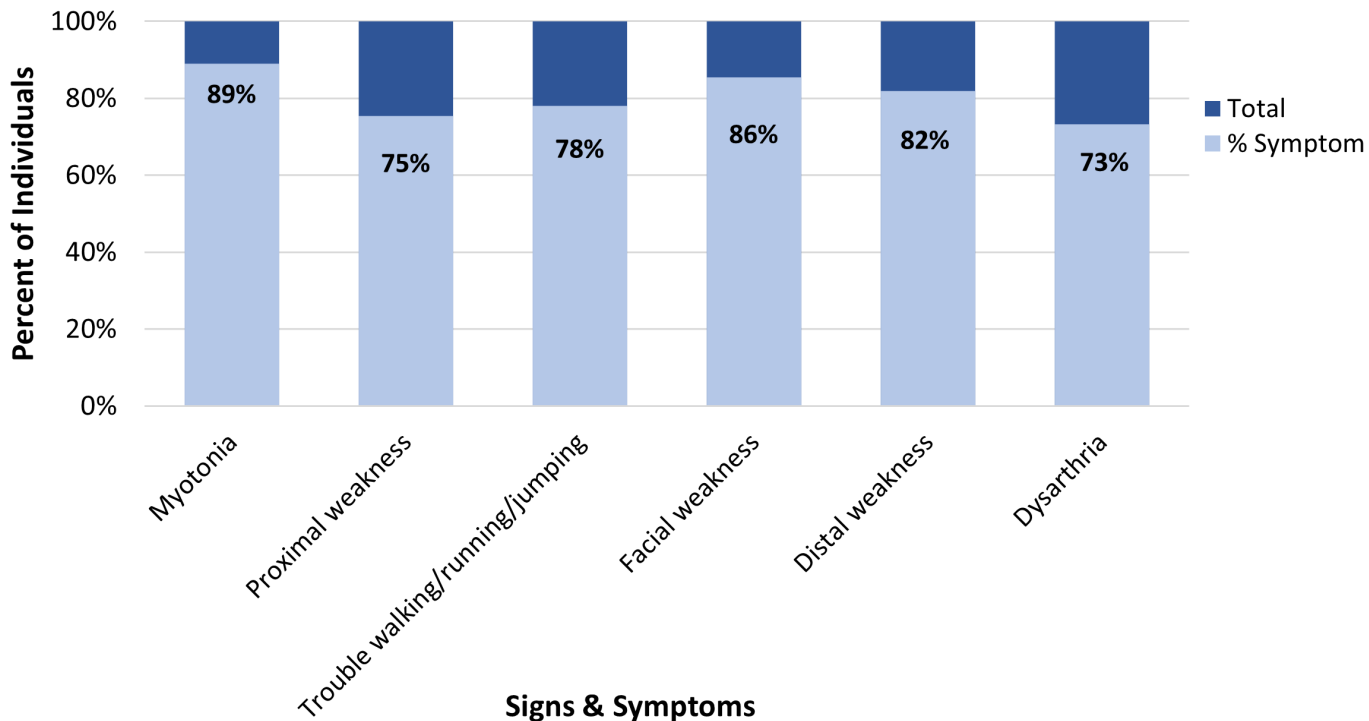
Figure 9c. Mobility Status of Individuals Diagnosed with Myotonic Muscular Dystrophy Not Otherwise Specified

Percent of Individuals (y-axis)

| Mobility	Percent
Independent Ambulation	48%
Assisted Ambulation	19%
Wheelchair (full-time)	23%
Wheelchair (part-time)	3%
Ambulation (unknown)	6%
Not Walking	0
Wheelchair (unknown)	0

Mobility (x-axis)

There were 31 individuals diagnosed with Myotonic Muscular Dystrophy that was not otherwise specified as of their last clinic visit. Almost half of the individuals (48%) reported independent ambulation, followed by those who were wheelchair-dependent full-time (23%), those who reported assisted ambulation (19%), and those who used a wheelchair part-time (3%).

Figure 10a. Signs and Symptoms in Individuals Diagnosed with Myotonic Muscular Dystrophy Type 1

Percent of Individuals (y-axis)

Signs & Symptoms	% Symptom
Myotonia	89%
Proximal weakness	75%
Trouble walking/running/jumping	78%
Facial weakness	86%
Distal weakness	82%
Dysarthria	73%

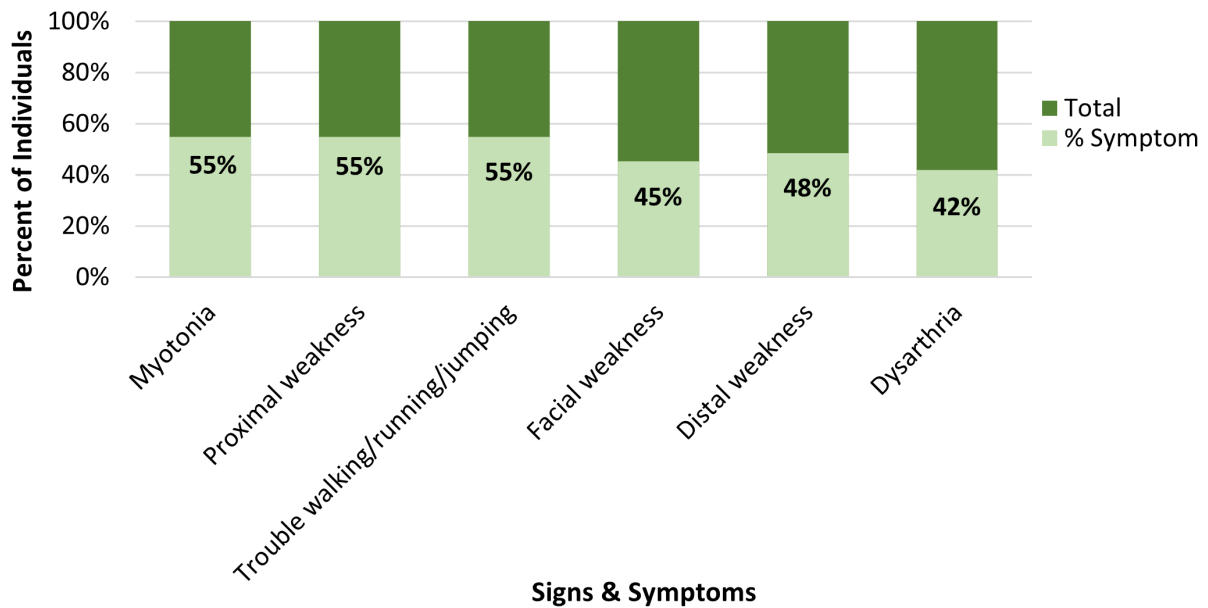Legend: ■ Total ■ % Symptom

Signs & Symptoms (x-axis)

Individuals may be included in more than one of these categories. Each category represents the proportion of individuals out of the total (n=228). The graph represents the most frequent symptoms that individuals experienced based on medical records. Myotonia, facial weakness, and distal weakness were the most frequently reported signs/symptoms in individuals diagnosed with Myotonic Muscular Dystrophy Type 1.

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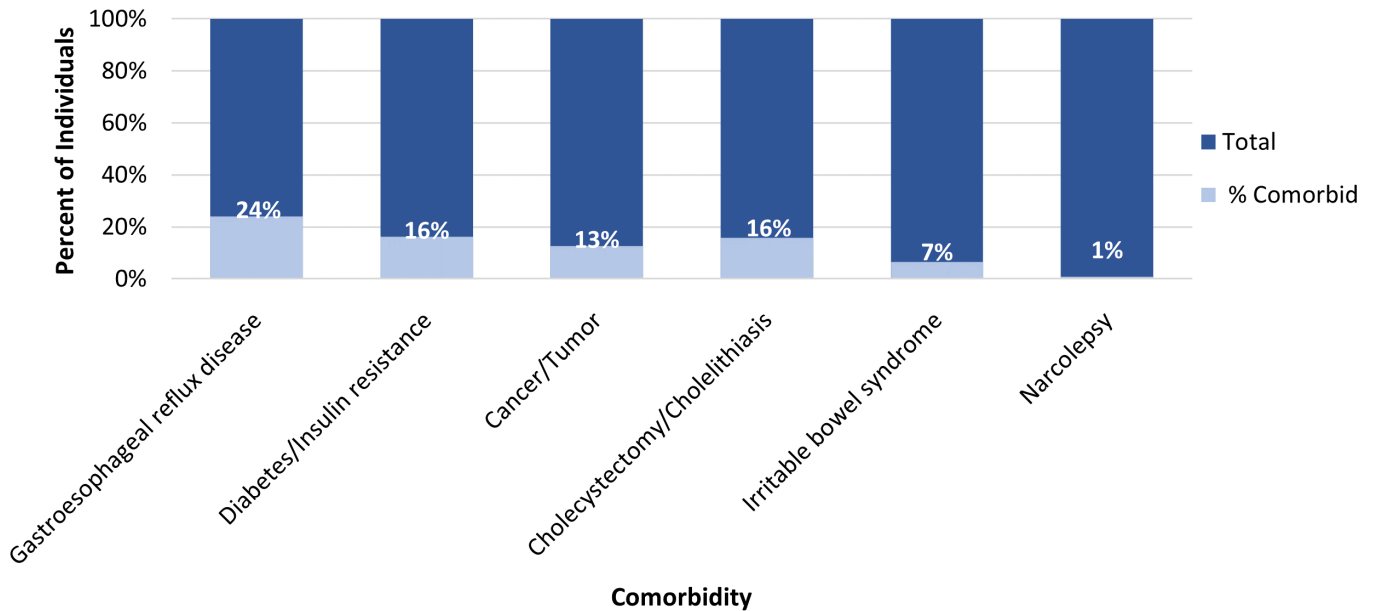## Figure 10b. Signs and Symptoms in Individuals Diagnosed with Myotonic Muscular Dystrophy Type 2
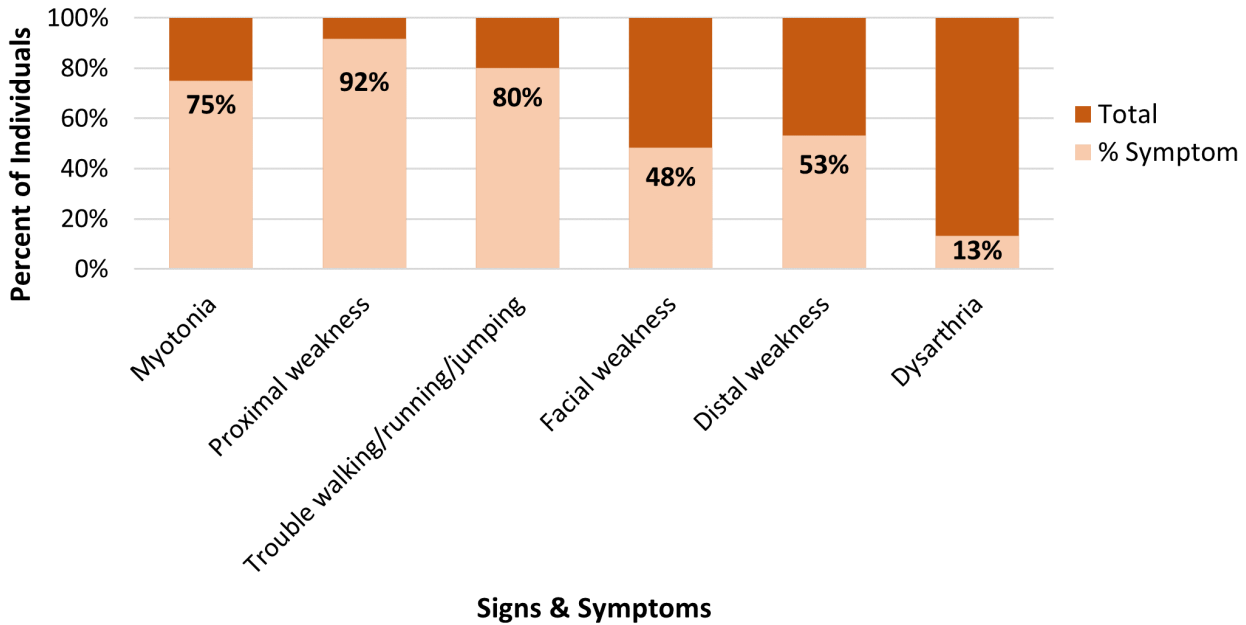
Each category represents the proportion of individuals out of the total (n=60). Proximal weakness, trouble walking, running, jumping and myotonia were the most frequently reported signs/symptoms in individuals diagnosed with Myotonic Muscular Dystrophy Type 2.

Figure 10c. Signs and Symptoms in Individuals Diagnosed with Myotonic Muscular Dystrophy Not Otherwise Specified
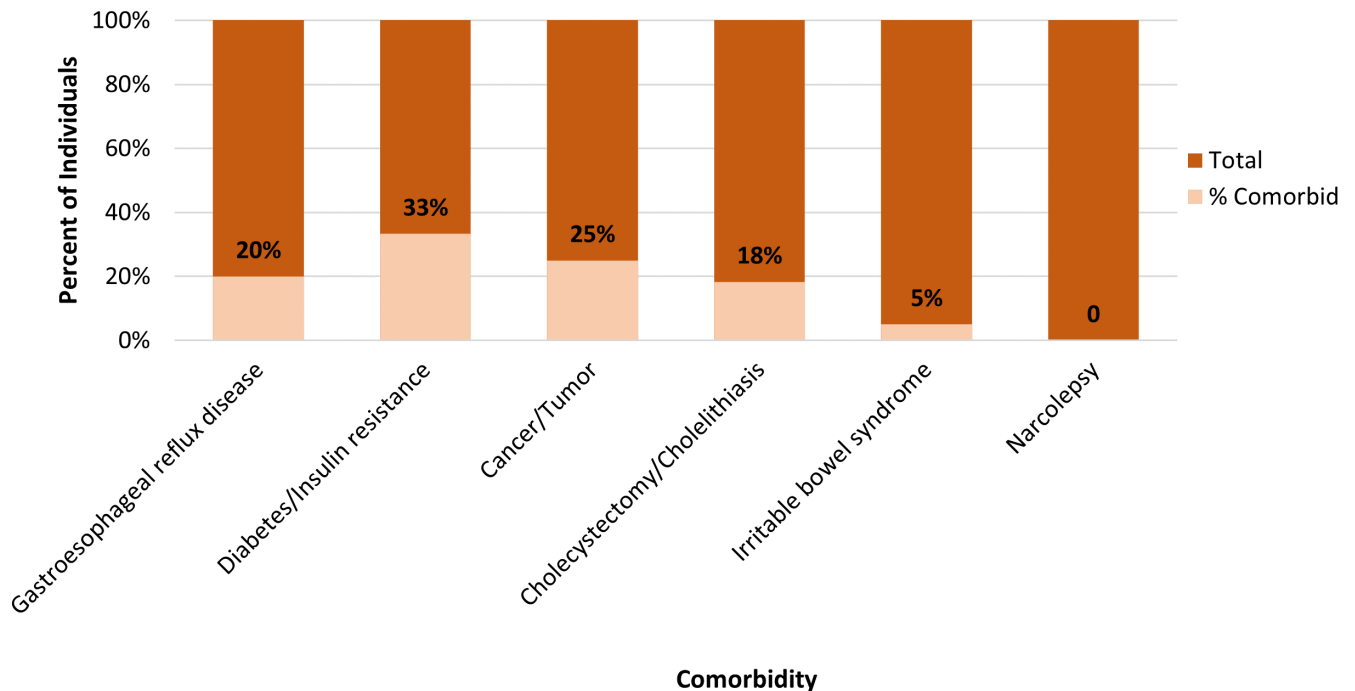

Each category represents the proportion of individuals out of the total (n=31). Family history, myotonia, proximal weakness, and trouble walking, running, jumping were the most frequently reported signs/symptoms in individuals diagnosed with Myotonic Muscular Dystrophy that was not otherwise specified.

Figure 11a. Comorbid Conditions in Individuals Diagnosed with Myotonic Muscular Dystrophy Type 1
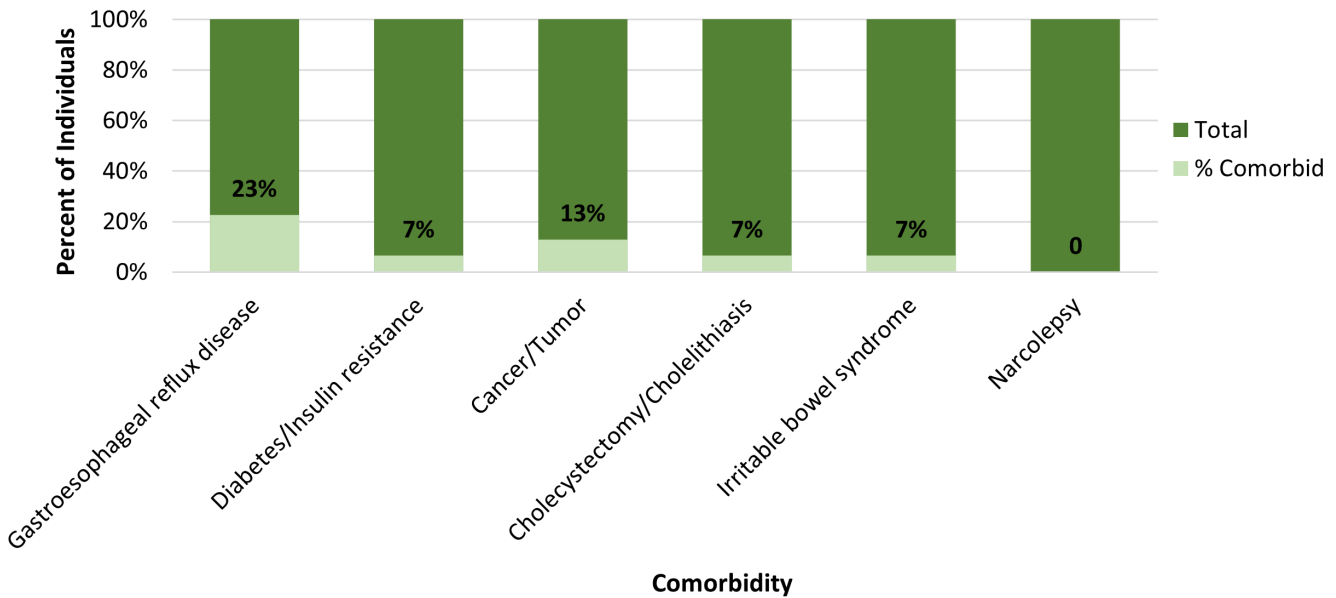

Individuals may be included in more than one of these categories. Each category represents the proportion of individuals out of the total (n=228). Gastroesophageal reflux disease was the most frequently reported comorbidity, while narcolepsy was the least commonly reported in individuals diagnosed with Myotonic Muscular Dystrophy Type 1.

Figure 11b. Comorbid Conditions in Individuals Diagnosed with Myotonic Muscular Dystrophy Type 2

Each category represents the proportion of individuals out of the total (n=60). Diabetes/ insulin resistance was the most frequently reported comorbidity, followed by cancer/tumor in individuals diagnosed with Myotonic Muscular Dystrophy Type 2.

Figure 11c. Comorbid Conditions in Individuals Diagnosed with Myotonic Muscular Dystrophy Not Otherwise Specified

Each category represents the proportion of individuals out of the total (n=31). Gastroesophageal reflux disease was the most frequently reported comorbidity, followed by cancer/tumor in individuals diagnosed with Myotonic Muscular Dystrophy that was not otherwise specified.
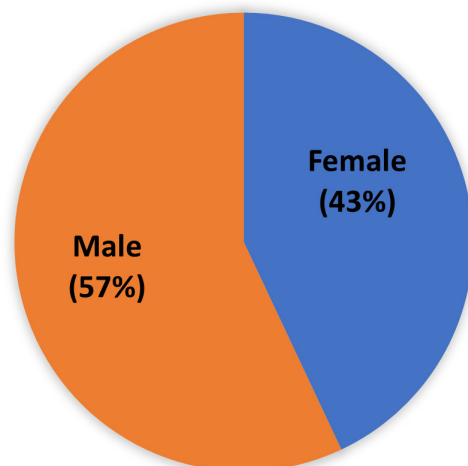
Facioscapulohumeral Muscular Dystrophy

The following data are based on a cohort of 107 individuals who were diagnosed with Facioscapulohumeral Muscular Dystrophy in Western NY between 1/1/2008 and 1/1/2016, and who had at least one healthcare visit on or after 1/1/2008. The start date of 1/1/2008 allowed for the most comprehensive and complete record access. The data include sociodemographic characteristics such as sex, race/ethnicity, vital status, health insurance coverage, care needs, employment status, and educational attainment of individuals; and clinical characteristics such as distribution of Facioscapulohumeral Muscular Dystrophy type, proportion of individuals who have at least one parent diagnosed with Facioscapulohumeral Muscular Dystrophy, mobility status, signs and symptoms, and comorbidities among individuals with Facioscapulohumeral Muscular Dystrophy.
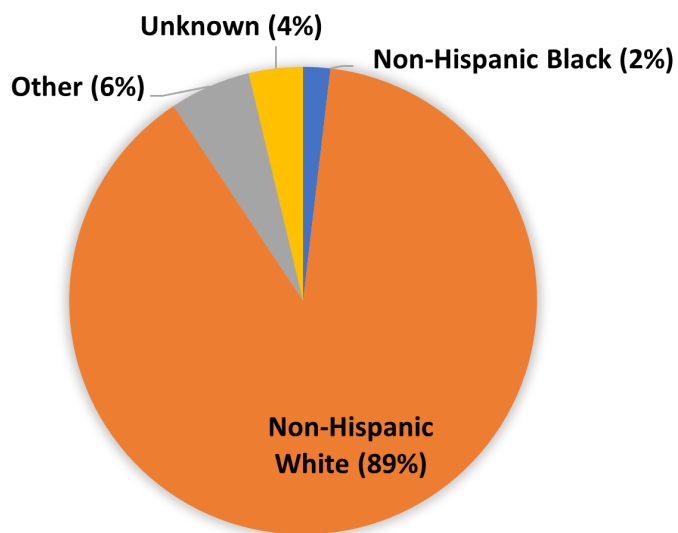
Sociodemographic Characteristics

Figure 12. Sex of Individuals Diagnosed with Facioscapulohumeral Muscular Dystrophy

More than half of the individuals in the cohort who were diagnosed with Facioscapulohumeral Muscular Dystrophy were males (57%).

Figure 13. Race/Ethnicity of Individuals Diagnosed with Facioscapulohumeral Muscular Dystrophy

Unknown (4%)

Other (6%)

Non-Hispanic Black (2%)

Non-Hispanic White (89%)

The majority of individuals identified as Non-Hispanic White (89%). Information about race was not available for 4% of the cohort.

Figure 14. Vital Status of Individuals Enrolled in the Study with Facioscapulohumeral Muscular Dystrophy

Deceased (5%)

Living (95%)
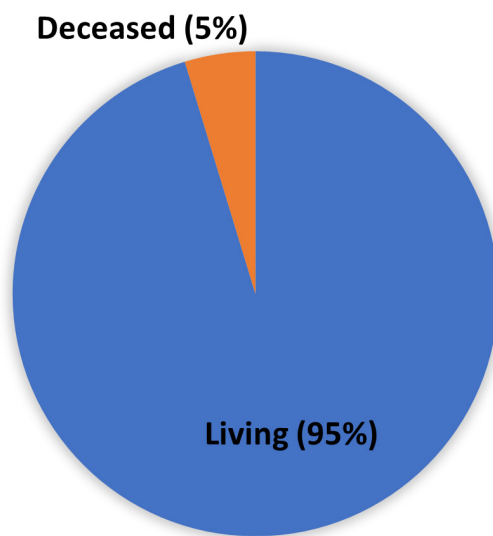
Most of the individuals in the cohort were alive (95%) at the time of data collection.

Figure 15. Health Insurance Coverage for Individuals Diagnosed with Facioscapulohumeral Muscular Dystrophy
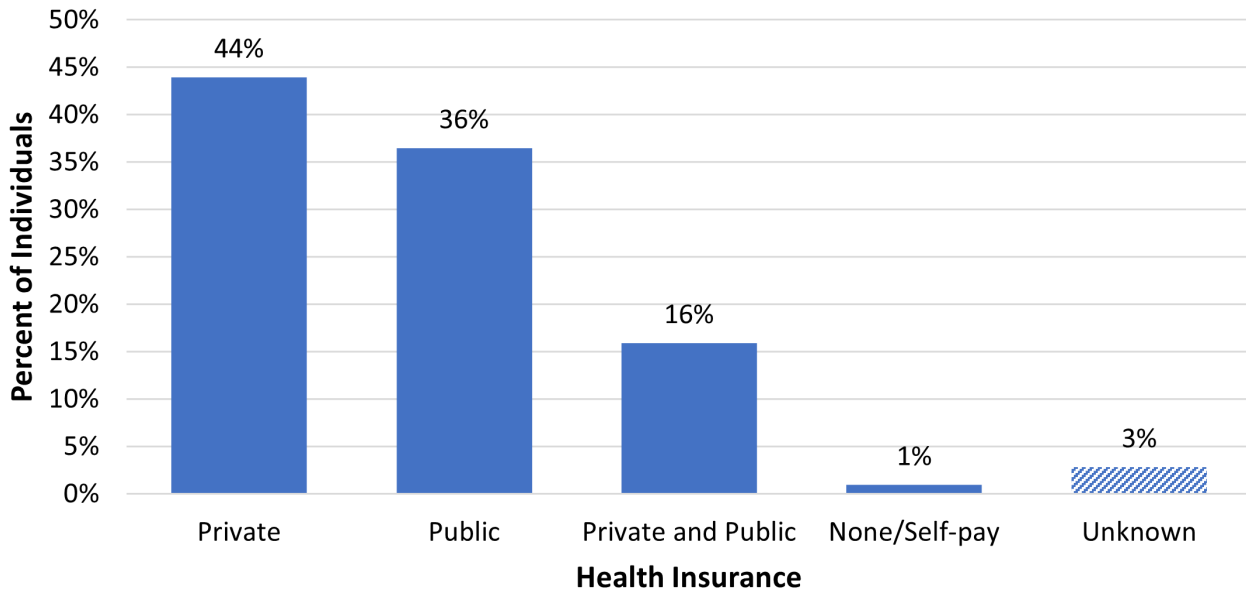

A higher proportion of individuals (44%) reported having private health insurance compared to other types of health insurance, followed by public (36%), then both public and private (16%). A small percentage (1%) of individuals reported other means of funding.

Figure 16. Care Need of Individuals Diagnosed with Facioscapulohumeral Muscular Dystrophy
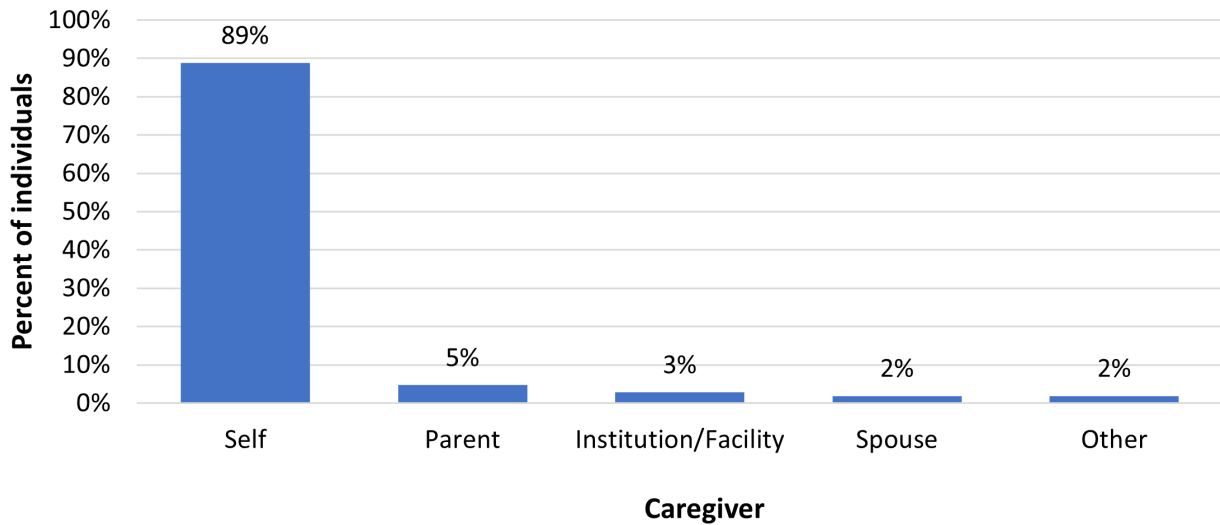
Most individuals reported caring for themselves (89%), while approximately 5% were cared for by their parents.

Figure 17. Employment Status of Individuals Diagnosed with Facioscapulohumeral Muscular Dystrophy
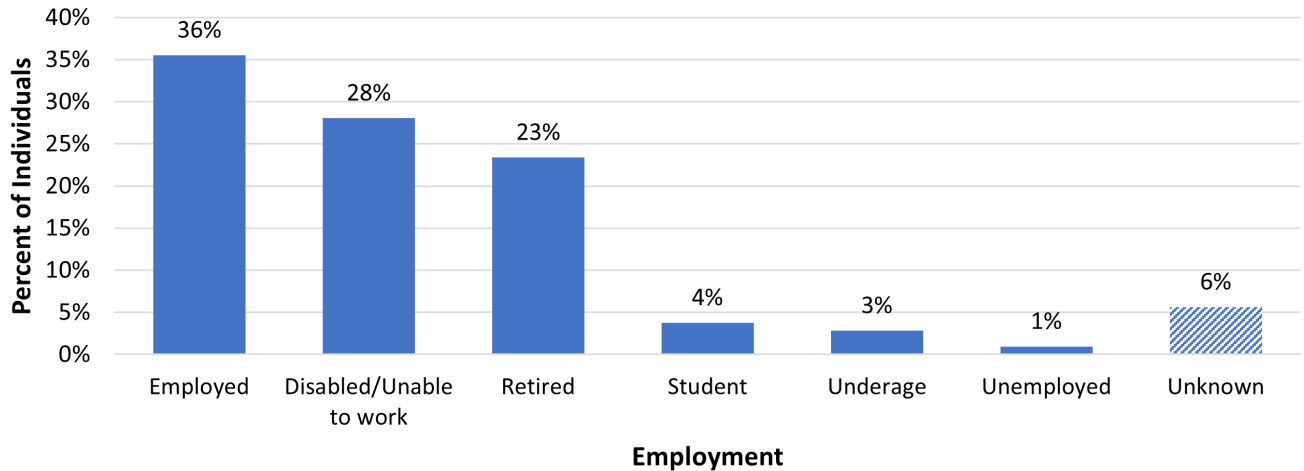
Approximately one third of individuals were employed (36%), followed by those who were disabled/unable to work (28%), and those who had retired (23%). The smallest percentages represent those who were students, underage, or unemployed. There was no employment status information for 6% of the individuals.

Clinical Characteristics

Table 2. Clinical Characteristics of Individuals Diagnosed with Facioscapulohumeral Muscular Dystrophy

Clinical Characteristics	Mean (Min-Max)
Age at last clinic visit (in years)	52 (13-88)
Age at partial or complete loss of mobility (in years)	62 (30-77)

Figure 18. Distribution of Facioscapulohumeral Muscular Dystrophy Type Among Individuals Diagnosed with Facioscapulohumeral Muscular Dystrophy

Most of the individuals had a Facioscapulohumeral Muscular Dystrophy Type 1 diagnosis. There was no Facioscapulohumeral Muscular Dystrophy type documented for 21% of the individuals.
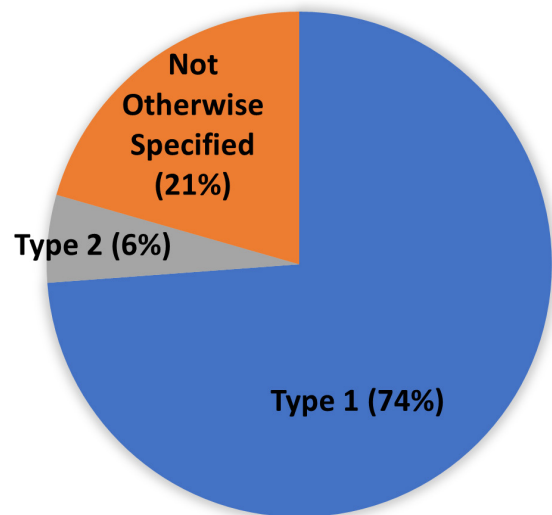
Figure 19. Mobility Status of Individuals Diagnosed with Facioscapulohumeral Muscular Dystrophy
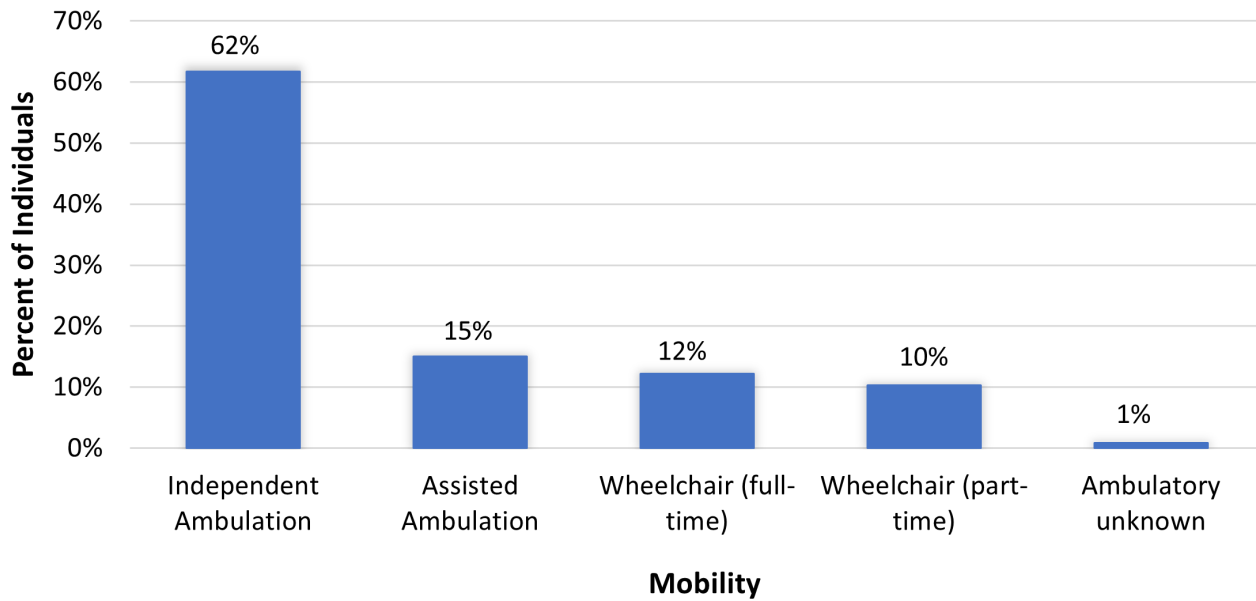
Most of the individuals (62%) reported independent ambulation, followed by those who reported assisted ambulation (15%), those who were wheelchair-dependent full-time (12%) and those who used a wheelchair part-time (10%).
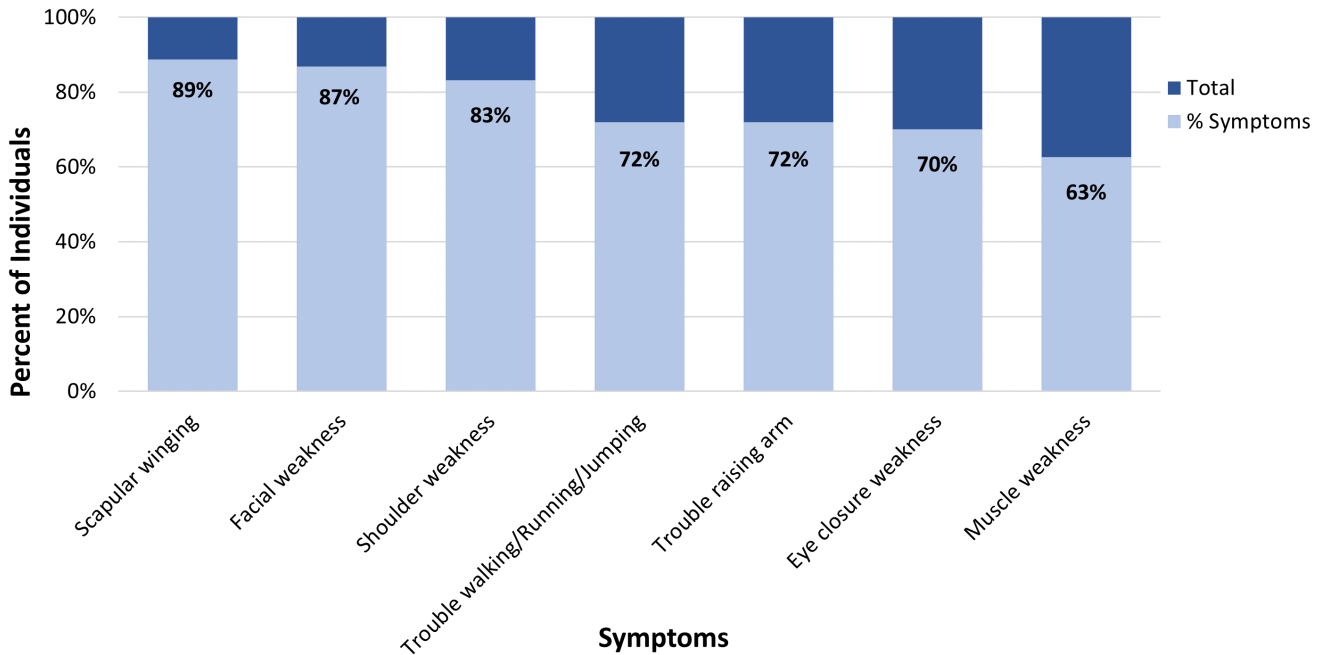
Figure 20. Proportion of Individuals Diagnosed with Facioscapulohumeral Muscular Dystrophy Who Had At Least One Parent Diagnosed with Facioscapulohumeral Muscular Dystrophy
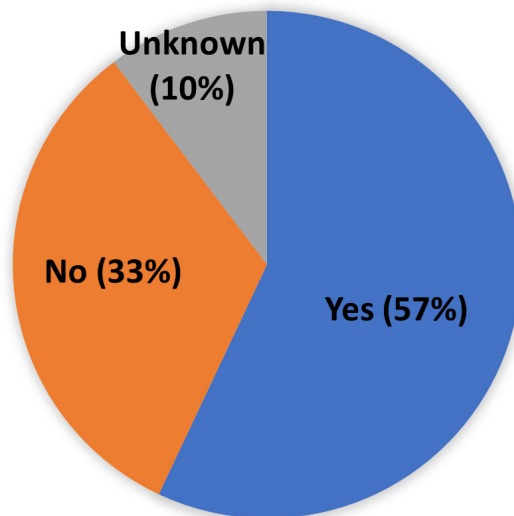
More than half of the individuals had at least one parent who also had a Facioscapulohumeral Muscular Dystrophy diagnosis. Information about parent diagnosis was not available for 10% of the cohort.
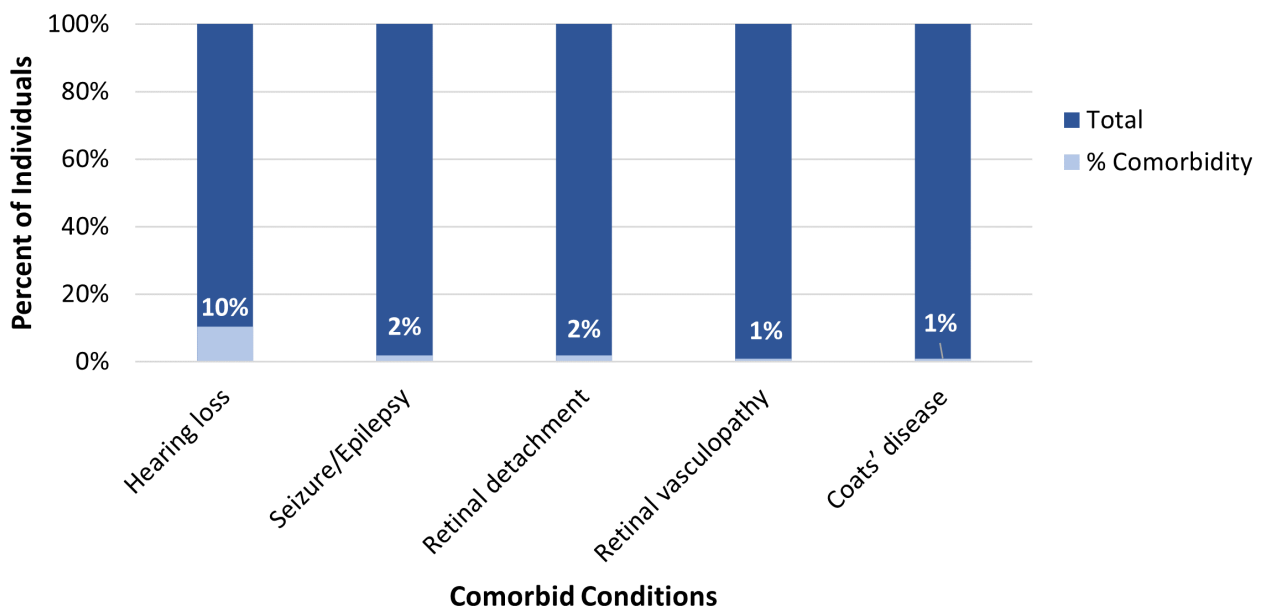
Figure 21. Symptoms of Individuals Diagnosed with Facioscapulohumeral Muscular Dystrophy

Individuals may be included in more than one of these categories. Each category represents the proportion of individuals out of the total (n=107). The graph represents the most frequent symptoms that individuals experienced based on medical records. Scapular winging, facial weakness and shoulder weakness were the most common symptoms reported in individuals diagnosed with Facioscapulohumeral Muscular Dystrophy.

**Figure 22. Comorbid Conditions in Individuals Diagnosed with Facioscapulohumeral Muscular Dystrophy**

Individuals may be included in more than one of these categories. Each category represents the proportion of individuals out of the total (n=107). The comorbid conditions were Coats' disease, epilepsy, hearing loss, retinal detachments and retinal vasculopathy. Of these, hearing loss was the most frequently reported (10%).

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Duchenne Muscular Dystrophy

The following data are based on a cohort of 87 individuals in Western NY who were born and diagnosed with Duchenne Muscular Dystrophy between 1982 and 2011. These data are based on their last clinic visit. The data include sociodemographic characteristics such as race/ethnicity, vital status, health insurance coverage, employment status, and educational attainment, and clinical characteristics such as distribution of mutation type, signs and symptoms, psychosocial conditions, medical interventions received, and therapy received among individuals with Duchenne Muscular Dystrophy.

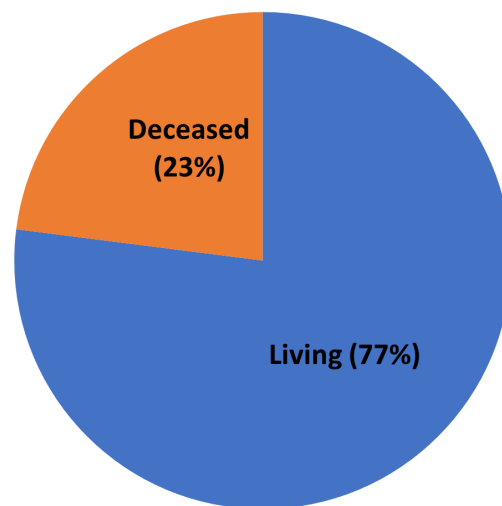## Sociodemographic Characteristics

Figure 23. Race/Ethnicity of Individuals Diagnosed with Duchenne Muscular Dystrophy
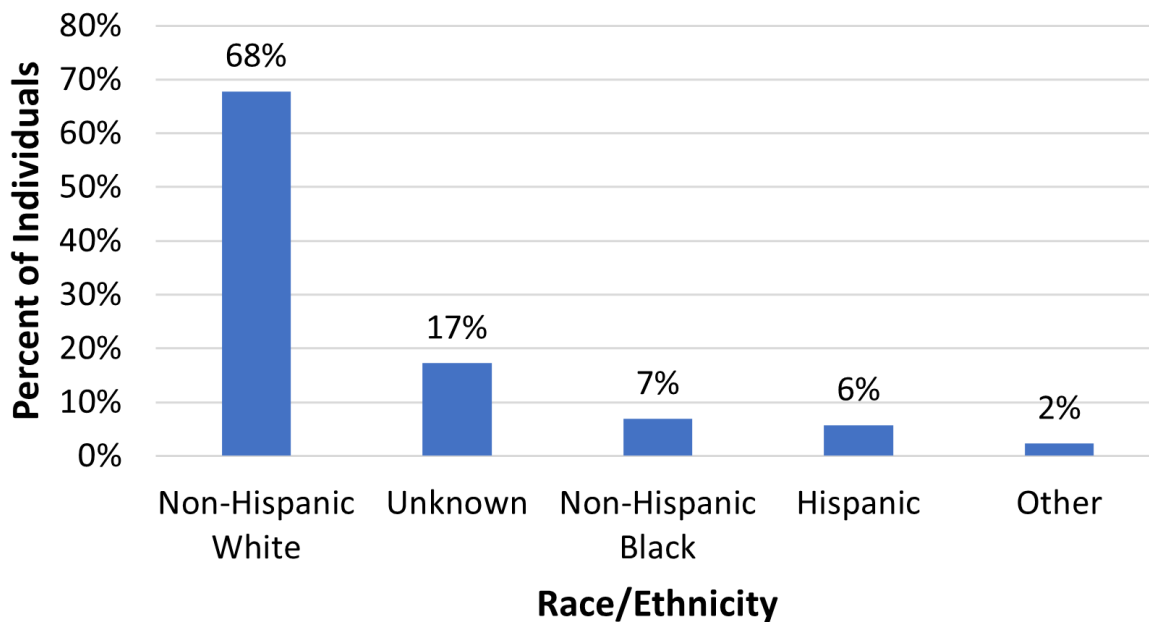

The majority of individuals diagnosed with Duchenne Muscular Dystrophy were Non-Hispanic White (68%). Information about race was not available for 17% of the cohort.

Figure 24. Vital Status of Individuals Enrolled in the Study with Duchenne Muscular Dystrophy

The majority of the individuals in the cohort were alive (77%) at the time of data collection.

Figure 25. Health Insurance Coverage of Individuals Diagnosed with Duchenne Muscular Dystrophy
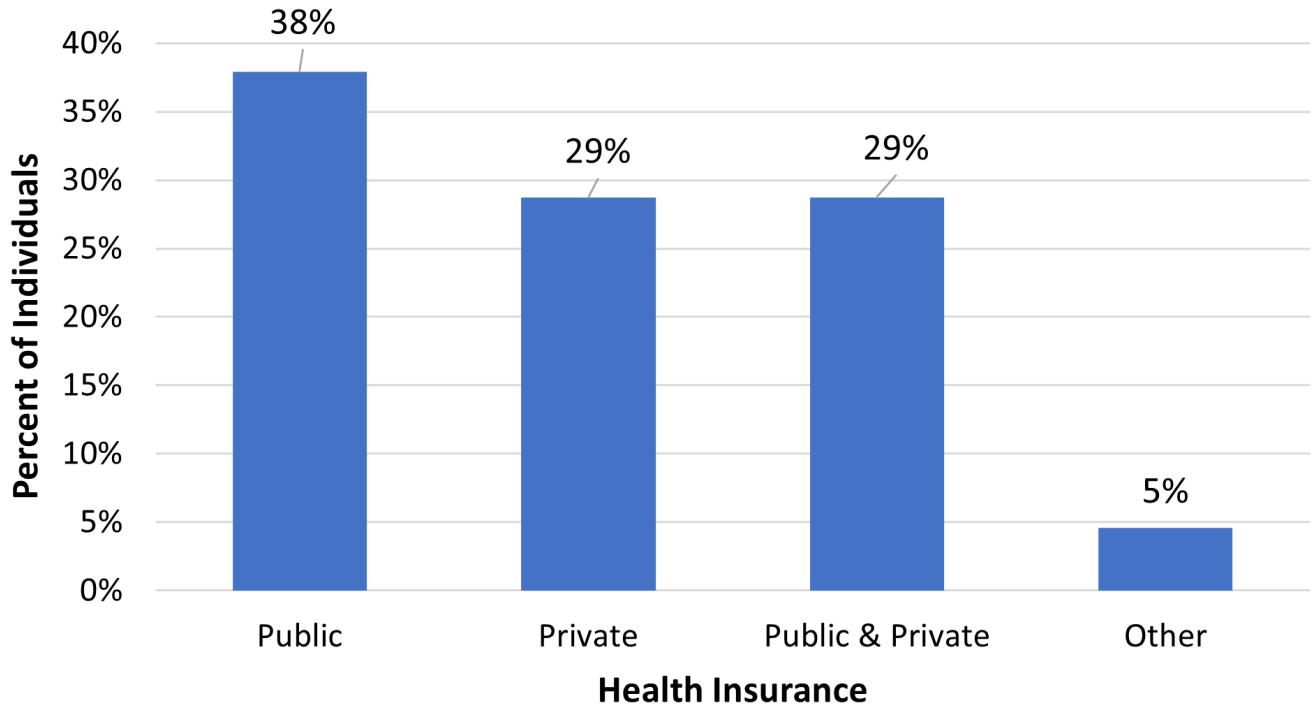

A higher proportion of individuals (38%) reported having public health insurance (Medicare and/or Medicaid) compared to other types of health insurance, followed by private (29%), and those who had both public and private (29%). A smaller percentage (5%) of individuals reported other means of funding.

Figure 26. Care Need of Individuals Diagnosed with Duchenne Muscular Dystrophy
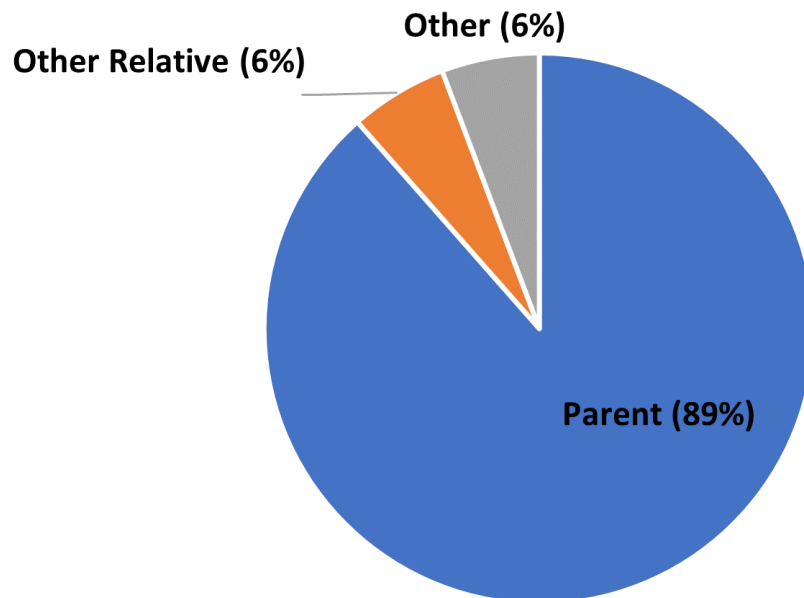

Most individuals (89%) received care assistance from their parents.

Figure 27. Employment Status of Individuals Diagnosed with Duchenne Muscular Dystrophy
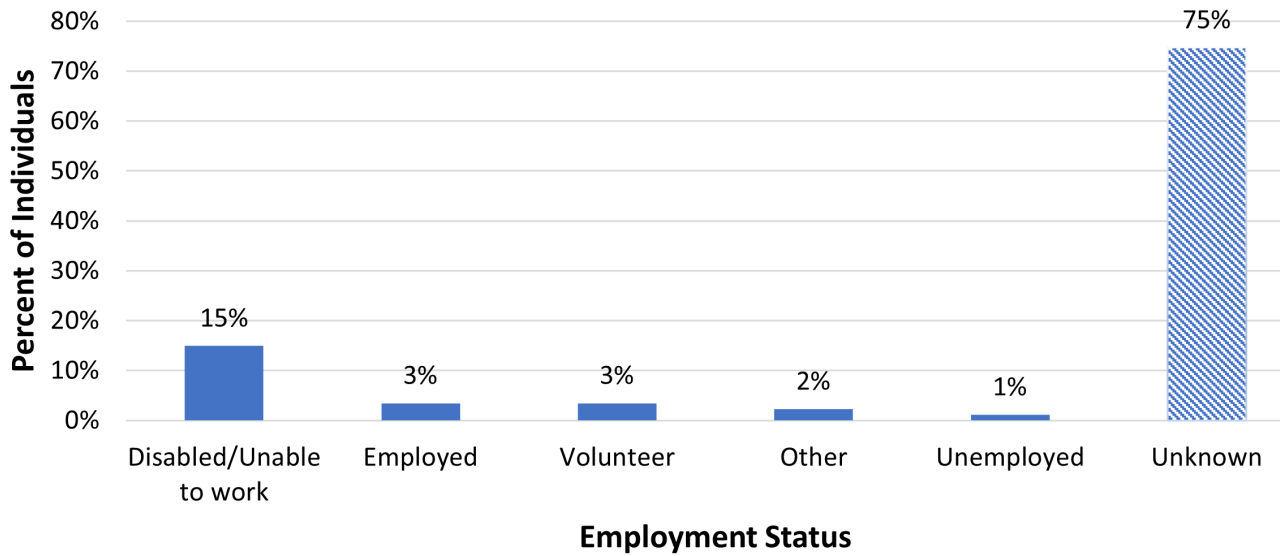
There was no employment information documented for most of the individuals (75%). Among those with employment information documented, most reported being disabled or unable to work, followed by those who were employed or did volunteer work.

Figure 28. Education Type of Individuals Diagnosed with Duchenne Muscular Dystrophy
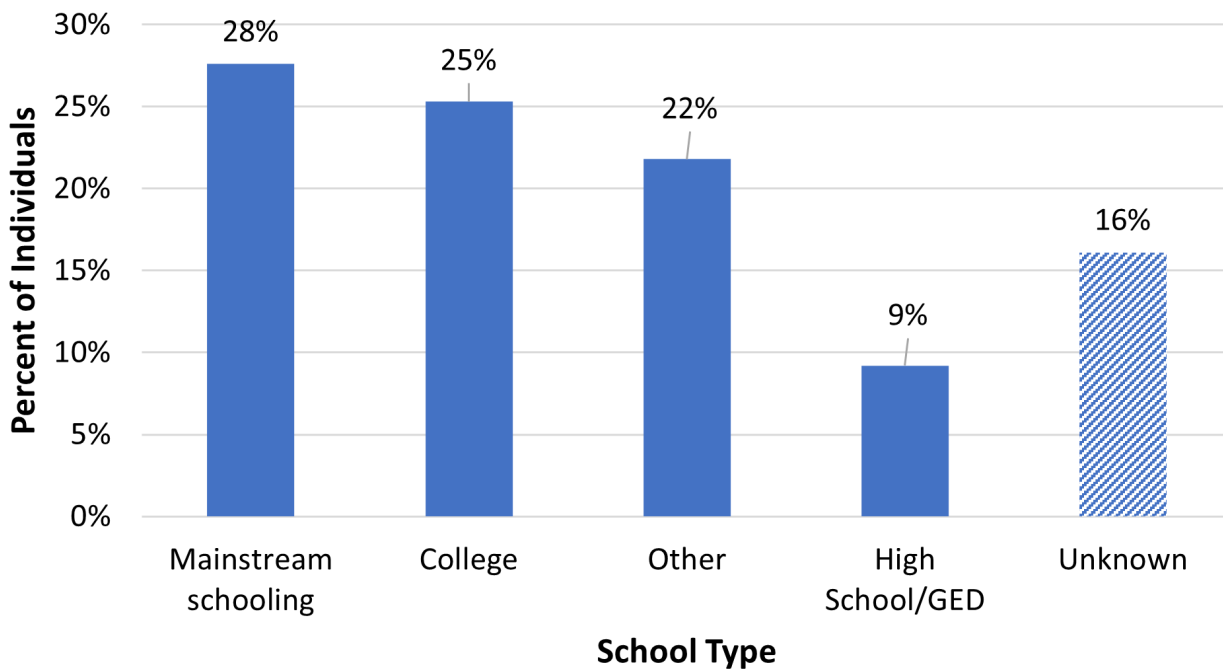

A higher proportion (28%) of individuals reported receiving mainstream schooling (in a general classroom), followed by college-level education (25%) and other types of schooling (22%), while 9% reported completing high school/GED. There was no school information documented for 16% of the individuals.

Clinical Characteristics

Table 3. Clinical Characteristics of Individuals Diagnosed with Duchenne Muscular Dystrophy

Clinical Characteristics	Mean (Min-Max)
Age at first abnormal cardiac echo (in years)	16 (5-24)
Age at first creatine kinase test (in years)	4 (0-16)
Age at first symptom (in years)	2 (0-8)
Age ambulation ceased (in years)	12 (6-23)
Age at death (in years)	21 (5-27)

Figure 29. Mutation Type for Individuals Diagnosed with Duchenne Muscular Dystrophy
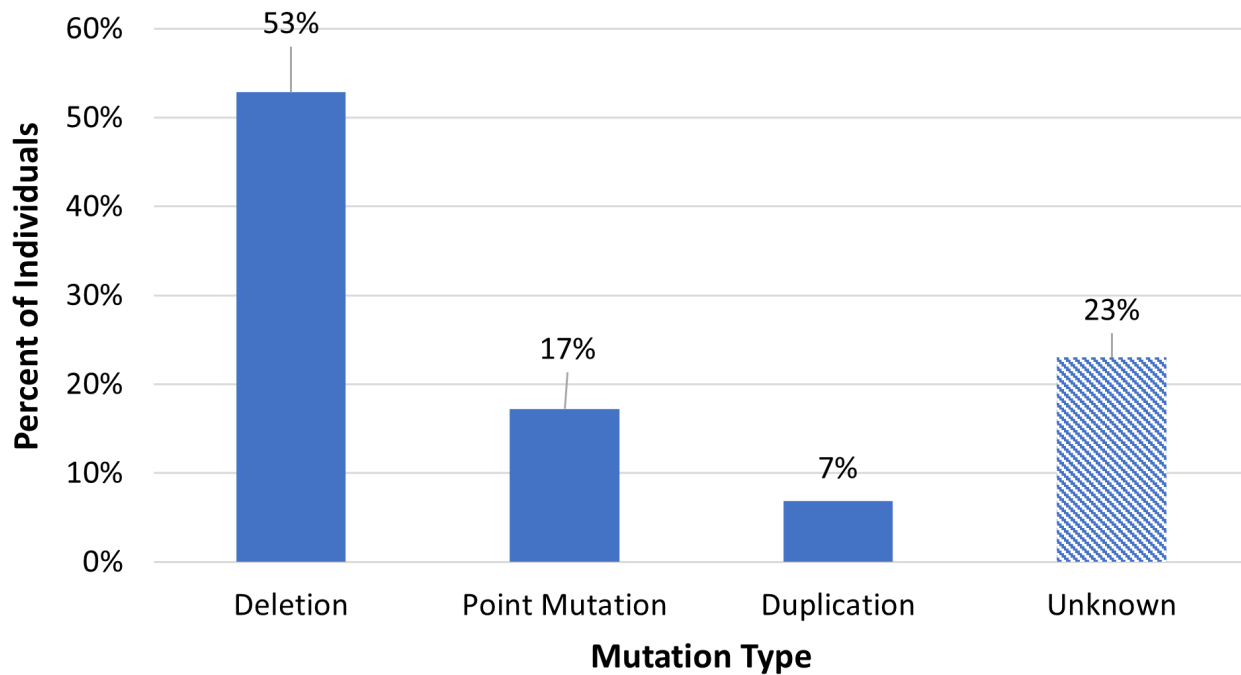
There was no information available on genetic mutation type for 23% of the individuals (n=20). More than half of the individuals had a deletion mutation type (53%), followed by a point mutation (17%). Only a small percentage (7%) had a duplication mutation type.
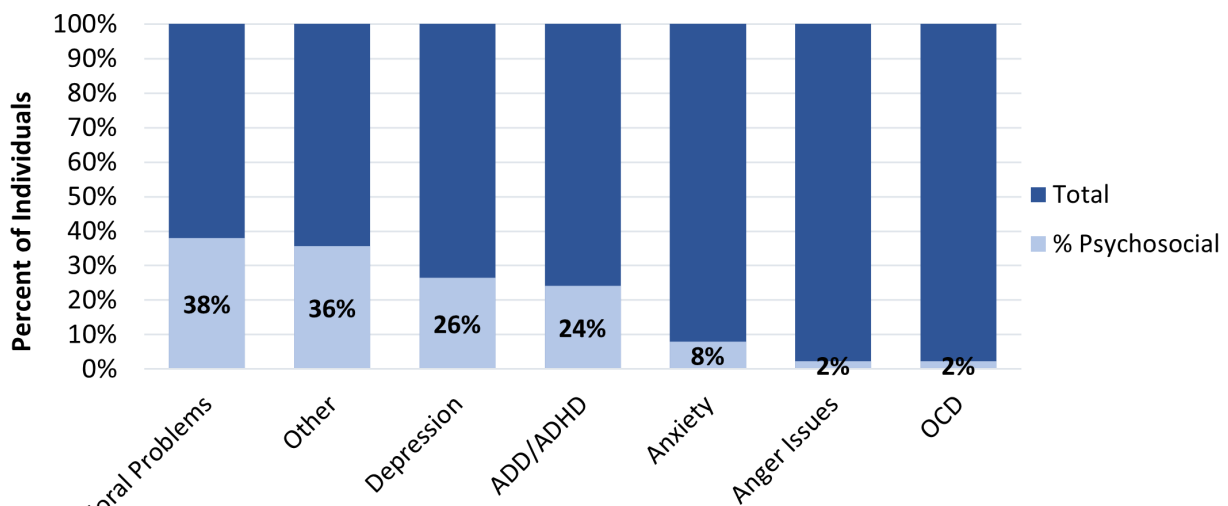
Figure 30. Signs and Symptoms of Individuals Diagnosed with Duchenne Muscular Dystrophy
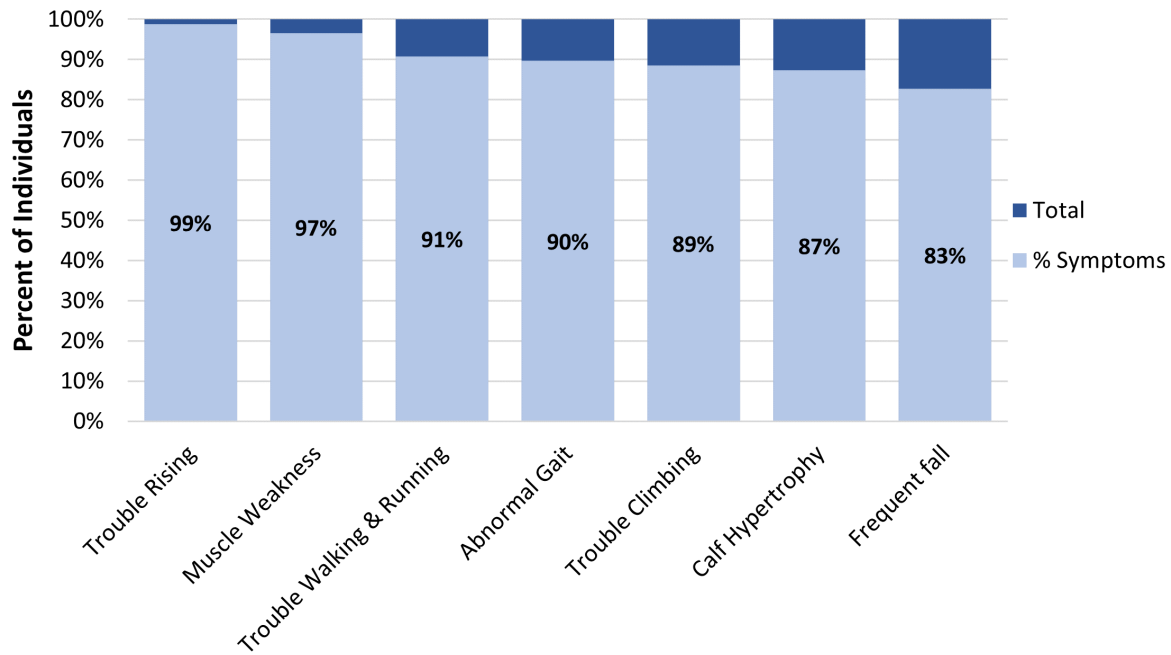
Individuals may be included in more than one of these categories. Each category represents the proportion of individuals out of the total (n= 87). The graph represents the most frequent symptoms that individuals experienced based on medical records. Almost all individuals diagnosed with Duchenne Muscular Dystrophy experienced trouble rising and muscle weakness.
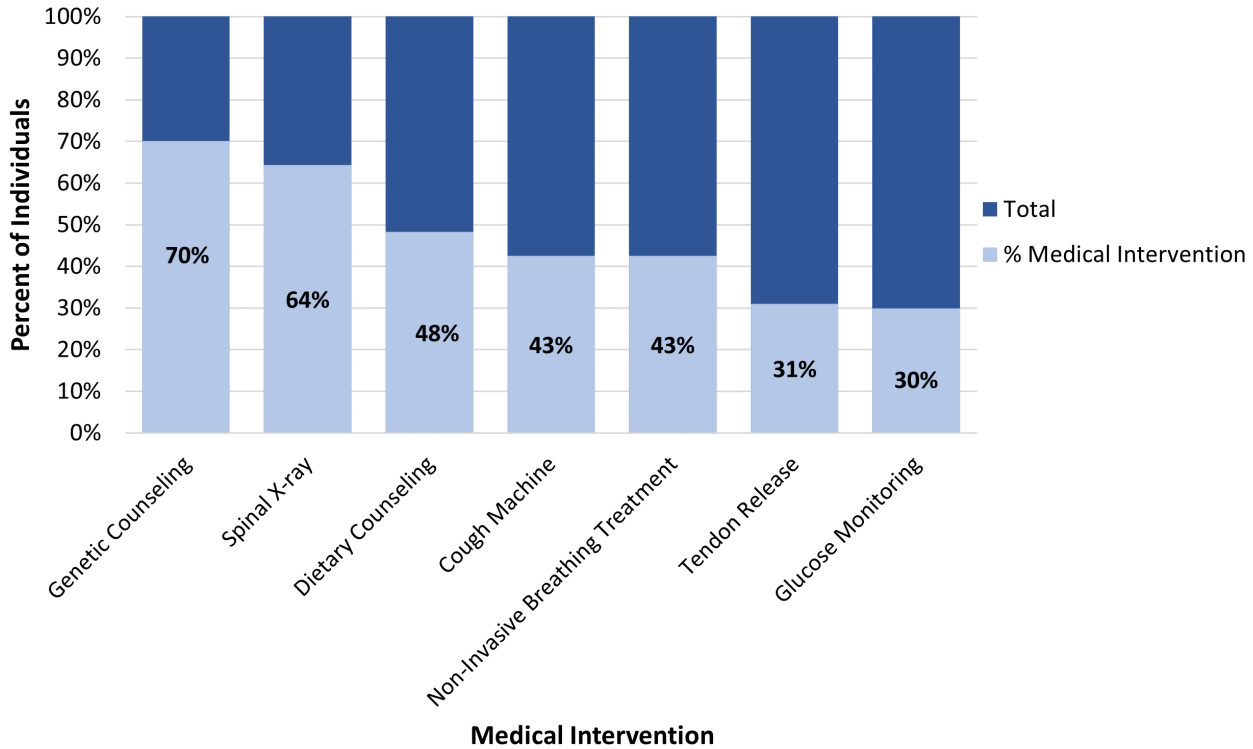
Figure 31. Psychosocial Conditions of Individuals Diagnosed with Duchenne Muscular Dystrophy

Individuals may be included in more than one of these categories. Each category represents the proportion of individuals out of the total (n= 87). A greater proportion of individuals reported behavioral problems (38%), other psychosocial issues (36%), depression (26%) and attention deficit disorder (ADD) or attention deficit hyperactivity disorder (ADHD) (24%).

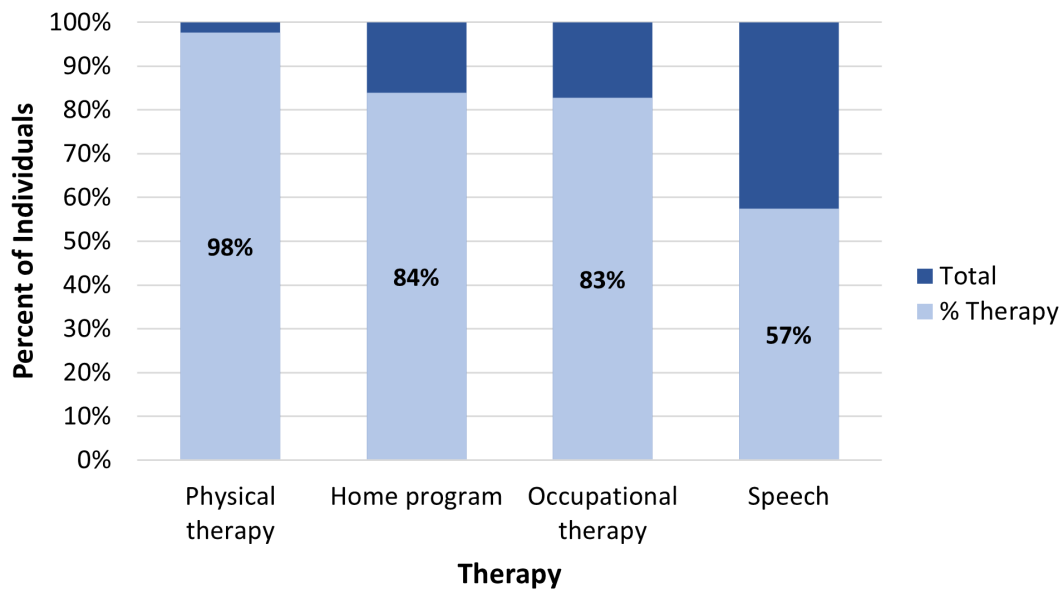# Figure 32. Medical Intervention Received by Individuals Diagnosed with Duchenne Muscular Dystrophy

Individuals may be included in more than one of these categories. Each category represents the proportion of individuals out of the total (n=87). A greater proportion of individuals received genetic counseling (70%), spinal x-ray (64%) and dietary counseling (48%).

Figure 33. Therapy Received by Individuals Diagnosed with Duchenne Muscular Dystrophy

Individuals may be included in more than one of these categories. Each category represents the proportion of individuals out of the total (n=87). Almost all individuals received physical therapy. A high proportion also received home program therapy and occupational therapy.

Summary

MD STAR*net* is the only research program designed to monitor and collect data for those diagnosed with muscular dystrophies who live in specific areas of the United States. By collecting these data, the MD STAR*net* program yields broader evidence-based research to help improve the care and quality of life for those living with muscular dystrophies. Population-based surveillance provides important information about the number of people affected by a disease, treatments received, and characteristics of affected people for the development of targeted interventions. The collection of clinical information allows for studies about natural histories and the impact of treatments on disease progression. The potential impact of surveillance data includes:

- Informing drug development companies about the potential population that would benefit from a new medication

- Informing clinical trial design about the number of potential participants

- Informing policy makers about the impact of muscular dystrophies on a population basis

- Informing Food and Drug Administration (FDA) rules by classifying muscular dystrophies as "rare diseases"

- Informing resource materials for families and individuals affected by muscular dystrophies, based on identified needs

Questions?

Email the New York MD STAR*net* team at **bdls@health.ny.gov**.

Learn more about MD STAR*net* at **www.cdc.gov/ncbddd/musculardystrophy/research.html.**