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Summary of Selected Healthcare Encounters among a Selection of Patients with Myotonic Muscular Dystrophy

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Abstract

Objective—Research has not examined the use of health care by patients with myotonic muscular dystrophy (MMD), but it would provide insights into this population, which is prone to comorbidities and high service needs. This study is an analysis of this understudied subgroup, using a unique linked dataset to examine the characteristics and healthcare utilization patterns for people with MMD.

Methods—This analysis used 3 South Carolina datasets (2009–2014). The subjects included individuals with at least 1 encounter with an *International Classification of Diseases, Ninth Revision, Clinical Modification* code of 359.21. The variables included sex, race, visit type, payer, and diagnoses. The analyses examined characteristics and number of encounters.

Results—The subjects were predominately female, white, and 45 to 64 years old. A total of 44.6% of the study population had at least 1 inpatient visit, whereas 64.2% had at least 1 emergency department visit. A majority of the subjects had at least 1 office visit (55.0%), and most (85.3%) did not have a home health encounter.

Conclusions—Investigation of the reasons for these inpatient and emergency department encounters may be helpful in identifying ways to deliver high-quality care.

Keywords

disability; emergency department; hospitalization; myotonic muscular dystrophy

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Muscular dystrophy (MD) can be any one of >30 genetic conditions, characterized by varying degrees of muscular weakness and movement disorders. Depending upon the specific disorder, the time of onset can range from infancy and childhood through to adulthood. Myotonic muscular dystrophy (MMD) is the most common adult-onset variant and is characterized by muscle control disorders, cardiac issues, and other abnormalities.[1]

MMD incorporates two broad types, myotonic dystrophy type 1 (DM1, previously referred to as Steinert disease) and myotonic dystrophy type 2 (DM2).[2] Both conditions are transmitted in an autosomal dominant pattern, although the affected chromosomes differ for DM1 versus DM2. Overall prevalence of myotonic dystrophy is estimated to be approximately 1 in 8000, with DM1 believed to be more prevalent than DM2.[2,3]

DM1 is characterized by variable age of onset, ranging from congenital DM1 (present at birth), to childhood onset (between ages 1 and 10 years), classic onset (between ages 10 and 30 years), and mild/late onset (between ages 20 and 70 years). Signs and symptoms also vary in type and severity but typically include myotonia and distal weakness, although for those with mild/late onset disease, both can be mild.[3] Additional clinical findings can affect an array of body systems and can include cardiac conduction abnormalities, cataracts, respiratory insufficiency, fatigue, cognitive deficits, and endocrine abnormalities such as testicular failure.[3] The leading cause of death for people with DM1 is respiratory failure, followed by cardiac disease.[4]

DM2 is generally milder and has later onset than DM1.[2] Proximal muscles are primarily affected, and affected individuals typically retain the ability to walk. Respiratory failure is typically not a concern and life expectancy is typically not substantially reduced, although cardiac conduction defects, cataracts, and endocrine abnormalities are common.[2] Pain tends to be a prominent feature affecting the quality of life of individuals affected by DM2.[5,6] There also is evidence that individuals with myotonic dystrophy (without differentiating between DM1 and DM2) have increased risk of a variety of cancers.[7]

Prior research has examined the healthcare utilization of individuals with MD without focusing on those with MMD specifically. This work among people with MD is instructive, however, because indicators of what gaps in service use or access may exist for this population. For example, children with MD were found to be less likely to have a medical home, a personal provider, or family-centered care than other children with special needs; these differences, however, were mitigated when family income and other factors were taken into account.[8] Those with MD also have been found to have higher rates of inpatient stays and emergency department visits than those without.[9] Others have examined the healthcare utilization of teenagers and young adults with MD enrolled in Medicaid; these results focused on racial differences, finding that African Americans with MD have been associated with a lower use of outpatient and specialist services and a higher use of inpatient stays and emergency department visits,[10] as well as higher mortality rates.[11] Studies have also shown differences in disease progression, comorbidities, and outcomes by sex.[12]

Other studies have examined the impact of insurance coverage, finding it to be a significant factor. Although a higher proportion of those with a disability have insurance, those without

coverage are more likely to experience barriers to access.[13] Even individuals with coverage have a higher likelihood of having unmet healthcare needs.[14–20]

Many studies to date do not focus solely on MD or MMD, but rather a more inclusive definition of disabling conditions. Given the wide range of clinical symptoms, comorbidities, and need for healthcare services among those with MMD, it is important to understand the patterns of healthcare use for this population. As such, the purpose of this study was to conduct such an analysis on this underexamined subgroup, by using a unique linked dataset to examine the characteristics and healthcare utilization patterns for people with MMD.

Methods

Data Sources and Study Design

This analysis used a combination of three distinct data sources within the state of South Carolina to conduct a case series analysis of the target population. The first two were obtained from the Revenue and Fiscal Affairs (RFA) Office within the SC Budget and Control Board. The RFA is a neutral service entity that warehouses administrative data and is able to link individuals across multiple service providers, locations, and data sources. The first data source was a deidentified dataset comprising claims for patients enrolled in either the State Health Plan (SHP) or Medicaid (fee for service and managed care enrollees). The SHP is an insurance plan administered by Blue Cross/Blue Shield of South Carolina that is offered to state and local government employees and their dependents. The two claims sources were combined before the analysis, with individuals matched and unduplicated using an internal matching algorithm (hereafter referred to as the SHP/Medicaid dataset).

The second data source was deidentified administrative data obtained via the Uniform Billing Form-04 and Centers for Medicare & Medicaid Services-1500 billing system. These data include diagnostic information (*International Classification of Diseases, Ninth Revision, Clinical Modification* [*ICD-9-CM*]), types of services received (*ICD-9-CM* procedures or Healthcare Common Procedure Coding System/Current Procedural Terminology procedures), timing of services, patient disposition, and other visit information for all inpatient and emergency department visits in the state, regardless of payer (hereafter referred to as the all-payer dataset). The use of these data for research among those with rare conditions has been documented elsewhere.[21]

The third data source was a dataset obtained from the Clinical Data Warehouse (CDW), administered by Health Sciences South Carolina. The CDW aggregates deidentified electronic patient records from the seven largest hospital systems in the state, and includes data related to inpatient, outpatient, and emergency department encounters within these systems.

To ensure the privacy and confidentiality of the records, the data obtained from the Health Sciences South Carolina CDW were sent to the analysts within the RFA. The analysts then matched the patient records using a proprietary algorithm. This algorithm relies upon matching identifiable fields, such as name, date of birth, and Social Security number (not

released to the public). Each record is given a numeric score based upon the number of fields matched. This process was iterative, with a minimum required score to identify a match. These matches are then assigned the same unique identifier. The process can account for misspellings, name changes, and slight differences in the date of birth. Once assigned, the individual's unique identifier remains associated with all of the subsequent episodes of services in data captured by RFA, regardless of data source or service provider. The unique identifier was then used to match patients across the three datasets and to ensure that records were not duplicated when the sets were merged. A detailed description of the combined dataset is presented elsewhere.[22]

Subjects were initially included in the analysis if they had at least 1 encounter in any of the 3 datasets, coded with an *ICD-9-CM* code of 359.0 (congenital hereditary muscular dystrophy), 359.1 (hereditary progressive muscular dystrophy), or 359.21 (myotonic muscular dystrophy). Diagnosis of MMD (both DM1 and DM2) typically relies upon a combination of history and clinical findings, clinical testing (eg, electromyography), and DNA testing.[2,3,23] Unfortunately, diagnosis codes in the *ICD-9* and *ICD-10* do not distinguish between DM1 and DM2. Thus, for this analysis, patients with MMD were identified as those with at least 1 encounter of any type with an *ICD-9-CM* code of 359.21. The data encompassed the period of January 2009 through August 2014.

Variables

The variables that were included in this analysis were patient sex, race (white, black, other), visit type (emergency department, inpatient, outpatient), payer (commercial, government, self-pay, other), timing of services, and diagnoses associated with the visit. Outpatient visits were further divided into office visits, home health, auxiliary services (including case management, transportation, or other support services), and other. These variables were present in each of the three data sources, and encounters were included in the final combined dataset, as described elsewhere.[22] The primary reason for a visit was categorized using the Clinical Classification Software system developed by the Healthcare Cost and Utilization Project.[24]

Statistical Analysis

The initial analysis first examined the proportion of MD cases that also had a diagnosis of MMD and then the study population and its characteristics. Estimates of the number of encounters per person and encounter type also were calculated on a per-annum basis. Because of prior work indicating that sex and age affect the expression of MMD, the analysis also was subset by sex to identify differences in healthcare utilization based on these characteristics.[25] Differences by groups, such as sex, were tested using A Wald χ^2 test. The sex subanalysis frequencies by diagnostic category were calculated by service type.

Results

The initial sample included 1801 individuals with MD. Of these, 307 (17.0%) had MMD. Table 1 displays the frequency of MD and MMD diagnoses within this study population, with marked differences across the sources. Within the UB data, having an MD or MMD

diagnosis documented in >1 encounter was relatively rare, with approximately one-third having >1 occurrence. These proportions were higher for the SHP/Medicaid data, where nearly 3 of 4 had >1 MD diagnosis occurrence, and nearly 4 of 5 had >1 MMD diagnosis occurrence. The CDW had the highest rate, with >90%having >1 MD or MMD diagnosis occurrence, respectively. There was a low proportion (<20%) with an MMD diagnoses and a concurrent MD diagnosis in all of the datasets.

Table 2 summarizes the characteristics of the MMD study population. The MMD cohort was predominately female and white. Because the SHP/Medicaid data suppress race information, nearly one-third of the sample was missing a race designation. The largest age group was 45 to 64 years, followed by 25 to 44 years and 0 to 19 years. The age distribution differed by sex: a larger proportion of males than females were in the 0 to 19 age group, and a lower proportion of males were in the 20 to 44 age group compared with females (Table 3). Table 4 displays the proportion with each encounter type during the entire study period, subset by sex. Overall, 44.6% of the study population had at least 1 inpatient visit, whereas 64.2% had at least 1 emergency department visit. A majority of the encounters were in the outpatient/other category, with only 7.5% of the population not having any encounters of this type. Of these, a majority had at least 1 office visit (55.0%), whereas larger majorities did not have a home health (85.3%) or auxiliary (86.6%) encounter. The only significant difference by sex was for receipt of auxiliary services; a smaller proportion of females (7.7%) had these encounters than males (20.3%, $P < 0.05$).

Table 5 displays the estimates of healthcare encounters, by type, on a per-annum basis. Among those with MMD, the average number of inpatient stays per year was 0.33 and emergency department encounters averaged 0.68/year. There were no significant differences by sex for these two encounter types.

The outpatient/other encounter category displays greater frequency and variability because of the all-encompassing nature of the encounters contained. It does, however, give an indication of the frequency of contact with the healthcare system, because it includes ambulatory physician visits, outpatient hospital encounters, and various other encounters. Outpatient encounters averaged 22.3/year, with males having more on average (27.8) than females (17.7, $P < 0.05$). Differences also were observed by sex in the types of outpatient/other visits. It is not surprising that office visits accounted for the largest proportion of this type, with an average of 7.8/year. For office visits, males had nearly twice as many on average (10.1) than females (6.0, $P < 0.05$). Those with MMD averaged >7 home health encounters and nearly 2 auxiliary services per year. These encounter types did not differ by sex.

Tables 6A–C display the most common clinical classifications, by service categories and sex (Supplemental Digital Content, http://links.lww.com/SMJ/A149). For inpatient visits, the most common reasons included endocrine disorders (primarily *ICD-9-CM* 255: disorders of the adrenal glands), upper respiratory infections (primarily *ICD-9-CM* 472, chronic pharyngitis and nasopharyngitis), and pneumonia (primarily *ICD-9-CM* 486, pneumonia, organism unspecified). Neoplasms, injuries, and lung diseases were among the 10 most frequent diagnoses documented for inpatient visits among males, whereas the most frequent

diagnoses documented for inpatient visits among females included pneumonia and perinatal conditions.

For emergency department visits, the most frequent reasons for a visit included upper respiratory conditions (primarily *ICD-9-CM* 786 inclusive: symptoms involving respiratory system and other chest symptoms), back problems (primarily *ICD-9-CM* 724 inclusive: other and unspecified disorders of the back), and sprains and strains (primarily including *ICD-9-CM* codes 847 [back], 845 [ankle and foot], and 844 [knee and leg] inclusive). The conditions included in the upper respiratory category represent a broad range of concerns, including conditions of the nose and oropharynx, as well as some conditions of the larynx, trachea, and bronchi. The list of conditions in Clinical Classification Software category 205 ("spondylosis, intervertebral disc disorders, and back problems," in Table 6A) is a full range of spinal conditions, including specific conditions such as degenerative disc disease and spondylosis and less specific conditions such as back pain not otherwise specified. Conditions included in this category range from the cervical spine to the coccyx. These differed somewhat by sex: for emergency department visits, sprains and strains and upper respiratory disease were documented more frequently relative to other diagnoses in the top 10 diagnoses list for females than for males.

For all outpatient services combined, the most common diagnoses were developmental disorders (primarily *ICD-9-CM* codes 315, 317, 318, and 319 inclusive, which are used for developmental delays and varying severities of intellectual disability), nervous system disorders (primarily *ICD-9-CM* 359: muscular dystrophies and other myopathies), and behavioral issues (primarily *ICD-9-CM* 314: hyperkinetic syndromes of childhood, which includes attention-deficit/hyperactivity disorder and related conditions). Category 259 (residual codes, unclassified) also was prominent and primarily comprised *ICD-9-CM* codes relating to general symptoms (780 inclusive); symptoms concerning nutrition, metabolism, and development (783 inclusive); symptoms involving nervous and musculoskeletal systems (781 inclusive); organic sleep disorders (327 inclusive); and symptoms involving the head and neck (784). For all outpatient services combined, skin ulcers were more frequent diagnoses relative to other top 10 diagnoses for males; among females, abdominal pain and diabetes were more frequent diagnoses in the top 10 list.

Subset to office visits with a physician, the most common diagnoses were similar to the overall outpatient visits, with developmental disorders, nervous system disorders, behavioral issues, and mood disorders being the most common. For outpatient visits, cardiac issues and skin ulcer diagnoses were more frequent among the 10 most frequent diagnoses among males, whereas upper respiratory issues and diabetes mellitus were more frequent among the top 10 diagnoses for females.

Discussion

This analysis used a unique, combined dataset to better describe the characteristics and healthcare utilization patterns of patients with MMD, a rare disabling condition. This study population demonstrated several interesting findings regarding both demographics and healthcare utilization patterns. Notably, individuals with MMD had high rates of use for the

encounters studied, such as inpatient and emergency department visits. These encounters were relatively frequent, even though these patients also had a high number of other encounters such as office visits. These healthcare utilization patterns are certainly related to their condition, which requires more frequent medical care. These findings also mirror prior work within more general MD populations.[20,21,23]

Several important differences by sex were identified within this study population; females were slightly overrepresented in this population (55% of the study population) and were more likely to be in the young adult age range. This may be caused by differences in access to insurance, particularly as patients age out of Medicaid coverage obtained as children. These data do not, however, allow for a full analysis of insurance coverage or changes in such coverage throughout the life span.

In addition, females had a lower proportion in the 19 and younger age group and a higher proportion in the 65 and older age groups. Typically, it would be expected that older populations would have higher healthcare utilization rates. These results indicated that a larger proportion of females did have at least one inpatient visit and also were more likely to make an emergency department or office visit. Females used auxiliary and home health services at a lower rate, perhaps offset by the higher use of other services. Examining the relation among sex, age, and types of healthcare utilization in this population may help determine whether the higher use of high-cost services (eg, inpatient visits, emergency department visits) among females in particular is warranted or potentially avoidable.

Those with MMD in this study also demonstrated a relatively high use of inpatient and emergency department services. On average, patients had one to two such encounters each year; among those with such an encounter, the frequency per year was higher still (2–3 inpatient or emergency department encounters per year, on average; data not shown). This utilization is despite frequent office visits (12–18 visits per person per year) and other services among this group. These findings do mirror other studies on individuals with MD[8–10]; additional research could help explain the reasons for these inpatient and emergency department encounters among those with MD and MMD to help decrease the reliance on these higher-cost encounters, where possible.

These findings indicate the potential value of coordinated care programs in such a population. With high healthcare utilization overall, particularly costly emergency department and inpatient stays, such programs could shift care to the outpatient setting, as appropriate. These programs have been studied extensively in other disease populations and have proven effective at reducing avoidable hospitalizations, improving care, and reducing costs.[26–28] Additional research needs to be conducted that is related to the potential for care management approaches to reduce the need for emergency and inpatient care, and improve overall care, in this patient population.

Because of the relative rarity of MMD, it can be difficult to conduct robust studies. One initial hurdle for such work is that of identifying individuals with the condition using available data. Methods for identifying cases vary, but often include having more than one encounter coded with the diagnosis code of interest within a certain period or having an

inpatient stay with that diagnosis code. Because the diagnosis of MMD (both DM1 and DM2) relies upon a combination of history and clinical findings, clinical testing (eg, electromyography), and DNA testing that is not available in these data, we were unable to confirm that all patients with a diagnosis code for an encounter indicating MMD truly had the condition or whether it was used for diagnostic encounters meant to rule out the disease.[2,3,23] The fact that a large proportion of patients have more than one MMD diagnosis code, particularly in the electronic medical record–based data, is a good indicator that these are patients with the condition. This interpretation is further supported by the low rate of concurrent MD and MMD coding for the same patients, indicating a high likelihood of these patients having the MMD variant.

This work does include several limitations that are worth noting. The population included in this analysis has potential bias because it was drawn from individuals who had used health care at some point during the study period. Although those with MMD are frequent users of services, it is possible that there are those who did not use any healthcare services and are thus not included in the study. In addition, the reliance upon diagnosis codes and not clinical data raises the possibility that some patients identified as having MMD may in fact have other conditions, and the use of that *ICD-9* code is inappropriate. In addition, the data do not provide a method to verify whether the included patients have a confirmed diagnosis of MMD; this was mitigated by the inclusion criteria of more than one instance of the diagnosis code in the data, and has been verified elsewhere.[21] The sources of data also present limitations because they do not encompass all of the individuals with MMD in the state. These data sources do, however, have a strength in the capture of all billed encounters by those patients, particularly all inpatient and emergency department visits. Also, not having the ability to differentiate between DM1 and DM2 may bias the results in some unknown way. Finally, the relatively small sample size naturally results in wider confidence intervals around the estimates. Larger studies with greater statistical power would improve the precision of the estimates.

Conclusions

The data presented in this analysis help us to better understand an understudied population of patients with a rare disabling condition. The results highlight several areas of potential study, including healthcare utilization patterns and substitution of one type of service for another, the impact of insurance coverage changes in utilization and outcomes, and the long-term outcomes of care received. These results may inform other work and provide direction on future research on healthcare utilization, cost, and outcomes in this population.

Supplementary Material

Refer to Web version on PubMed Central for supplementary material.

Acknowledgments

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Key Points

- Overall, 44.6% of the study population had at least 1 inpatient visit, while 64.2% had at least 1 emergency department visit. The average number of inpatient stays per year was 0.33, while emergency department encounters averaged 0.68 per year
- A majority had at least 1 office visit (55.0%), while larger majorities did not have a home health (85.3%) or other (86.6%) encounter. Outpatient encounters averaged 22.3 per year, with males having more on average (27.8) than females (17.7, $P < 0.05$).
- The reasons for encounters varied widely by type, but commonly included endocrine disorders, respiratory problems, and developmental delays.

Table 1.

Distribution of diagnoses within the study population, by data source, 2009–2014

	UB, %	SHP/Medicaid, %	HSSC,%
MD more than once	35.1	72.5	92.5
MD and MMD	15.3	18.1	14.4
MMD more than once	37.6	78.9	92.1

The categories are not mutually exclusive. HSSC, Health Sciences South Carolina Clinical Data Warehouse data; ICD, *International Classification of Diseases; MD, muscular dystrophy identified by* ICD *codes 359.0, 359.1, and 359.21; MMD, myotonic muscular dystrophy identified by* ICD *code 359.21; SHP/Medicaid, combined State Health Plan and Medicaid data; UB, Uniform Billing and Centers for Medicare & Medicaid Services-1500 billing system data.*

Table 2.

Characteristics of the study population, MMD only, 2009–2014

	N	%
All	307	100
Sex		
Female	169	55.0
Male	138	45.0
Race/ethnicity		
White	179	58.3
African American/Hispanic/other[a]	30	9.8
Missing	98	31.9
Age group, y		
0–19	75	24.4
20–24	17	5.5
25–44	80	26.1
45–64	108	35.2
≥65	27	8.8

MMD, myotonic muscular dystrophy.

[a]Combined because of small cell sizes.

Table 3.

Distribution of age by sex, MMD only, 2009–2014

	Female[a]		Male	
Age group, y	**Frequency**	**%**	**Frequency**	**%**
0–19	30	17.8	45	32.6
25–44	66	39.0	31	22.4
45–64	57	33.7	51	37.0
≥65	16	9.5	11	8.0

Age groups 20–24 and 25–44 years were combined because of small cell sizes. MMD, myotonic muscular dystrophy.

[a]Significantly different from male, $\alpha = 0.05$.

Table 4.

Percentage distribution of the number of encounters during the study period by type and sex, MMD only, 2009–2014

	All			Female			Male		
	0	**1**	**≥2**	**0**	**1**	**≥2**	**0**	**1**	**≥2**
Inpatient	55.4	17.3	27.4	53.3[a]	16.6	30.2	58.0	18.1	23.9
Emergency department	35.8	17.6	46.6	35.5	17.2	47.3	36.2	18.1	45.7
Outpatient	7.5	5.5	87.0	7.1	6.5	86.4	8.0	92.0[b]	
Office visit	45.0	55.0[b]		48.5[a]	51.5[b]		40.6	59.4[b]	
Auxiliary	86.6	4.6	8.8	92.3[a]	7.7[b]		79.7	7.2	13.0
Home health	85.3	14.7[b]		88.2[a]	0.0	11.8	81.9	18.1[b]	
Other	94.8	5.2[b]		95.9	4.1[b]		93.5	6.5[b]	
Unknown	3.6	7.5	88.9	3.0	4.7	92.3	4.3	10.9	84.8

MMD, myotonic muscular dystrophy.

[a]Significantly different from male, $\alpha = 0.05$.

[b]Categories combined because of small cell sizes.

Table 5.

Average number of annual encounters by encounter type and sex, MMD only, 2009–2014

	All		Female		Male	
Service	**Mean**	**SD**	**Mean**	**SD**	**Mean**	**SD**
Inpatient	0.33	0.91	0.32	0.82	0.34	1.01
Emergency department	0.68	1.45	0.78	1.63	0.54	1.19
Outpatient	22.25	47.56	17.71[a]	42.39	27.81	52.83
Office visit	7.84	15.71	5.99[a]	12.95	10.10	18.34
Auxiliary	1.87	12.50	1.02	6.64	2.92	17.12
Home health	7.02	29.72	7.38	32.49	6.58	26.03
Other	0.83	8.59	0.49	6.12	1.24	10.90
Unknown	5.70	19.31	3.94	8.24	7.85	27.21

MMD, myotonic muscular dystrophy; SD, standard deviation.

[a]Significantly different from male, $\alpha = 0.05$.