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Symptom Burden in Persons with Myotonic and Facioscapulohumeral Muscular Dystrophy

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Abstract

Objective—This study examines the prevalence of pain, fatigue, imbalance, memory impairment and vision loss in persons with myotonic and facioscapulohumeral dystrophy, and their association with functioning.

Design—A survey (n=170) included measures of severity (0–10 scales) and course of these symptoms, as well as measures of social integration, home competency, mental health and productive activity. Descriptive and regression analyses examined the associations between symptoms and functioning.

Results—Fatigue (91%), imbalance (82%) and pain (77%) were most commonly reported. The most severe symptom was fatigue (mean severity 5.14 ± 2.81), followed by imbalance (4.95 ± 3.25). Symptoms were most likely to stay the same or worsen since onset. Controlling for potential medical and demographic confounds, symptoms were associated with 17% of the mental health variance, 10% of home competency, 10% of social integration, 16% of productive activity for DM1 and 12% of productive activity for FSHD.

Conclusions—Pain, fatigue and imbalance are common in persons with muscular dystrophy. Interventions may be useful to mitigate their impact on functioning. Further research should examine these relationships to guide clinical practices.

Keywords

Neuromuscular Disease; Facioscapulohumeral Dystrophy; Myotonic Dystrophy; Patient Functioning

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Muscular dystrophy (MD) is a group of genetically distinct disorders characterized by progressive weakness and dystrophic changes in muscle, with loss of normal muscle fibers and replacement with fat and connective tissue. Myotonic dystrophy type 1 is a multi-systemic disease with common cognitive deficits in addition to the muscular symptoms. DM1 affects skeletal muscle, smooth muscle, myocardium, brain, and ocular structures, and is the only type of DM examined in the present study. Facioscapulohumeral muscular dystrophy (FSHD) is a slowly progressive dystrophic myopathy with predominant involvement of facial and shoulder girdle musculature. Prominent facial weakness is the hallmark of FSHD.

Although progressive muscle weakness is the hallmark symptom of MD, recent research indicates that many “secondary conditions”, such as chronic pain, fatigue, and imbalance, may also create significant burden for many persons with chronic neuromuscular disease. [1–4] These symptoms may also negatively impact quality of life (QoL) and patient functioning in this population. [5] We discuss each of these symptoms and its relevance in the sections that follow.

Pain in MD

Previous research has found that individuals with FSHD and DM1 experience a higher prevalence and greater severity of pain than do members of the general US population. Bushby and colleagues reported on four individuals with FSHD who identified pain as their most disabling symptom and complained of between three to seven separate pain complaints each. [6] Abresch and colleagues found that 83% of a sample of 811 individuals with various MDs, including 64 persons with FSHD and 33 with DM1, reported at least some ongoing pain problems. The frequency and severity of pain in their combined sample of patients with FSHD, DM1, and a sample of patients with limb-girdle syndrome was significantly greater than levels of pain reported by the general US population.[7]

Jensen et al surveyed 193 individuals with a variety of MDs, including 18 patients with FSHD and 26 patients with DM1, and found that 89% of patients with FSHD and 69% of those with DM1 reported pain problems. Severe pain was reported in 19% of patients with FSHD and 50% of patients with DM1. [8] Further, pain was reported to interfere with a number of activities of daily living (ADLs) [8]. Although the preliminary findings from our group and others indicate that chronic pain can be a serious problem for many persons with FSHD and DM1, much remains unknown about the nature and scope of pain in these patient populations. Moreover, because both FSHD and DM1 are progressive diseases, it is possible that the onset of pain and pain severity may be related to a patient’s age or degree of mobility impairment. However, these relationships have not yet been reported in the published literature.

Fatigue in MD

Fatigue is often present in patients with neurological diseases. Patients with FSHD and DM1 have been shown to not only report higher levels of fatigue than healthy controls, but those experiencing severe fatigue appear to have increased functional impairment [2,3,9,10]. Though many studies document the presence of fatigue in persons with MD, to our knowledge no

studies assess self-report of the course of fatigue and its tendency to resolve, worsen, or stay the same over time.

Imbalance in MD

Little is known about the prevalence of imbalance in persons with MD. Horlings and colleagues [11] reported that 72 persons with FSHD had a higher frequency of falls than healthy controls, and frequent fallers had lower measures of physical functioning. [11] Though other factors such as weakness and fear of falling may contribute to these findings, they also suggest the possibility that imbalance may be a significant concern for persons with FSHD as it may contribute to the prevalence of falls.

Memory Impairment in NMD

A few studies have reported that the prevalence of cognitive impairment, including impaired visuospatial memory, is higher in those with DM1 as compared to the normal population. [12,13] However, to our knowledge no studies have described the prevalence or course of self-reported memory loss in persons with DM1 or FSHD, or examined its tendency to interfere with functioning.

Vision loss in NMD

Little is known about the prevalence of vision loss in persons with DM1 or FSHD. However, some studies have shown that cataracts are more common in persons with DM1 and DM2 as compared to the general population. [13] These findings suggest further exploration into the prevalence of vision loss and its course over time.

Purpose of the Current Study

Research suggests that individuals with MD report problems with a number of symptoms, such as pain, fatigue, imbalance, memory problems, and vision loss. However, little is known regarding the relative frequency, course, and impact of these co-occurring symptoms in the same sample. In the present study, we sought to (1) determine the relative frequency and severity of these symptoms in a sample of persons with FSHD and DM1; (2) determine the extent to which these symptoms are reported to have a tendency to improve or resolve, get worse, or stay the same over time; and (3) examine the associations between the severity of these symptoms and measures of patient functioning in a sample of persons with FSHD and DM1.

Because each symptom could potentially play an important role in patient functioning, we hypothesized that the severity of each symptom would both (1) show significant zero order associations with study criterion variables and (2) show independent (i.e., when controlling for other symptoms and both chronological age and disability duration) associations with criterion variables controlling for other symptoms of patient functioning.

METHODS

Participants and Procedures

The participants for this study were recruited as part of a larger study examining chronic pain and disability in muscular dystrophy. [14] Participants in the survey came from two primary sources: (1) the National Registry of Myotonic Dystrophy and Facioscapulohumeral Muscular Dystrophy Patients and Family Members (http://www.urmc.rochester.edu/nihregistry) (n=296) funded by the National Institutes of Health and (2) the University of Washington Muscular Dystrophy Association (MDA) clinic roster (n=87). A small number of additional subjects were recruited from a previous study that they completed when they were children (n=8)[15]. An additional 4 participants heard about the study and independently contacted study personnel expressing their interest in participation.

Two hundred and seventy-two surveys were mailed to potential participants and 195 were returned, yielding a return rate of 83%. Data from 3 of these surveys could not be analyzed (because of insufficient data or determined ineligibility based on their survey responses) and were consequently excluded from further analysis, resulting in a sample of 192 participants. To control for potential confounds associated with the heterogeneity of this population, we narrowed down this sample to include only individuals with the two most common MD diagnoses (DM1 and FSHD), resulting in a final sample size of 171 participants (81 with DM1 and 90 with FSHD).

Each questionnaire was accompanied by a consent form and a cover letter inviting the potential participants to participate in the study. Subjects were paid $25 for completing and returning the consent forms and survey. The research methodology and all study procedures were approved by the University of Washington Human Subjects Review Committee.

Measures

The survey asked participants to provide basic demographic information (age, education level, employment status, race and ethnicity, marital status, and assistive device use) and information about their dystrophy diagnosis, including: type of dystrophy, method of diagnosis, and type of doctor who confirmed the diagnosis. Survey respondents were also asked about the presence, severity and course of five key symptoms (pain, fatigue, imbalance, memory impairment and vision loss). Severity of each symptom was assessed using a 0 (none) to 10 (very severe) Numerical Rating Scale (NRS), and symptom course was assessed by asking the participants to indicate, for each symptom, whether it had become worse, become better, or stayed the same since its onset, and during the past six months. All participants were also asked to complete the 13-item Community Integration Questionnaire (CIQ), which assesses participation in valued activities, including activities in the home (such as meal preparation and housework), social activities (such as leisure activities with others), and productive activity (such as employment). Evidence supports the reliability, discriminant validity and construct validity of the CIQ scales.[16,17]

Psychological functioning was measured using thee Medical Outcomes Study 36-Item Short Form Health Survey (SF-36) mental health scale, a measure with shown reliability and validity to assess four domains of mental health (vitality, social functioning, role-emotional

and mental health). [18] The scale is scored to have a possible range of 0–100, with better mental health indicated by higher scores.

Statistical Analyses

A symptom was defined as being present when a respondent rated its severity as being at least “1” on the 0–10 NRS. Average severity was computed for all participants who endorsed the symptom (i.e., rated the severity as being at least “1”). In order to determine the usual course of these symptoms, we computed the rate of each course type (worse, the same, better) associated with each symptom since onset and during the past 6 months.

In order to test the hypothesis that each symptom is independently associated with measures of psychological and physical functioning, we first performed regression analyses to determine if diagnostic group (FSHD versus DM1) had a moderating effect on the associations between symptom severity and the criterion variables. In the event of a significant moderation effect (indicating that the associations between symptom severity and a criterion variable differed between the two groups), we planned to perform two regression analyses – one using the DM1 participants and the second the FSHD participants – predicting the criterion variable. Otherwise, analyses were performed using both groups combined. The four measures of functioning were the criterion variables and the five symptom severity ratings (pain, fatigue, imbalance, memory loss, and vision loss) were the primary predictors in the analyses. Age and duration of MD were entered in the first step of these analyses in order to control for their possible confounding effects (e.g., because some symptoms and general disability might increase in frequency or severity with these age-related variables).

RESULTS

Participant Characteristics

All participants had a clinically confirmed diagnosis of FSHD or DM1. Most (61%) reported that they had a DNA confirmation of diagnosis, while others reported that they either had no DNA confirmation (24%) or that they didn’t know whether their diagnosis had been DNA confirmed (15%). The majority of participants (93%) were diagnosed by neurologists; other sources of diagnosis included other physicians such as NMD specialists, family practitioners, and physiatrists.

The mean age of the study subjects was 51.9 years (SD, 13.1 years; range, 21–90 years). There was a large variability in the number of years since neuromuscular disease diagnosis, with a mean of 18.9 years (SD, 11.8 years; range, 2.3–51.1 years). The majority (54%) of respondents were women. Participants were allowed to indicate more than one race/ethnicity classification. Most (168, or 99%) reported that they were white, 5 (3%) indicated that they were Hispanic (3%), and one participant (1%) reported an Asian or Pacific Islander background. The respondents reported variability in education level with 18% having a high school education or General Educational Development certificate, 8% having attended vocational or technical school, 23% having had some college, 33% being college graduates,

and 19% having attended graduate school. Only one person (1%) reported not graduating from high school.

Frequency of Symptoms

The most common symptoms reported by the survey respondents were fatigue (91%), imbalance (82%), and pain (77%) (see Table 1). The most severe symptom was fatigue, with 47% reporting fatigue levels of ≥ 7on the 0–10 scale [sample mean = 5.1 (2.8)]. 50% of the sample reported imbalance levels of ≥ 7 on the 0–10 scale, while pain (24%) and vision loss (12%) were reported as severe by a substantial subset of participants. Memory impairment was reported as less than severe, on average (4%).

Regarding course, all five of the symptoms that were assessed were more often reported as staying the same or getting worse rather than getting better since the onset of the symptom (see Table 2). During the past six months, symptoms tended most often to be perceived as staying about the same rather than getting worse or getting better.

Associations among Symptom Severity, Activity, and Psychosocial Function

Table 3 shows the zero-order correlation coefficients between the symptom severity ratings and the 4 measures of patient functioning. The results of these analyses show that symptoms reported by this sample were more closely linked to productive activity and psychological functioning than they were to social integration or home competency. The symptoms that were associated with all four domains (psychological functioning, social integration, home competency and productive activity) were vision loss and memory impairment. All statistically significant correlations were negative, indicating that the greater the symptom intensity, the lower the psychological functioning, social integration, home competency, or productive activity.

Testing for moderation effect revealed that dystrophy type moderated the effects of imbalance on productive activity. Thus, we performed three regression analyses predicting home competency, social integration and psychological functioning using a sample consisting of both diagnostic groups. Two regression analyses, one using the FSHD sample and the other the DM1 sample, were performed to predict productive activity. Controlling for age, sex, and duration of MD, the five symptoms examined as predictors were significantly associated with the CIQ social integration, home competency, and the SF-36 mental health scale in the combined sample (see Table 4). Symptoms were also significantly associated with productive activity in both diagnostic groups when analyzed separately (see Tables 5 and 6). Imbalance, though not a significant predictor on its own, was positively associated with productive activity in our FSHD sample, and negatively associated with productive activity in our DM1 sample. Further, memory loss was a significant negative predictor of productive activity in our FSHD sample, though not significant in our DM1 sample. Taken together, these symptoms accounted for 17% of the variance of the SF-36 Mental Health score, 10% of the CIQ Home Competency score, and 10% of the CIQ Social Integration score in the combined sample. Symptoms accounted for 16% of the variance in the CIQ Productive Activity score for DM1, and 12% of the CIQ Productive Activity score for FSHD over and above contributions of age, sex, and duration of MD. Age, sex, and

duration of MD did contribute significantly to the productive activity and home competency scales of the CIQ.

DISCUSSION

The study findings provide new information regarding the prevalence, severity, course, and impact of key secondary symptoms in persons with DM1 and FSHD. Consistent with the observations of clinicians who work with individuals with MD, fatigue and pain were very common. However, our results also underscore the importance of problems with balance in this population. Imbalance was not only one of the more prevalent symptoms in our sample of DM1 and FSHD, the majority of participants reported that their imbalance had worsened over the course of their disability. Imbalance can contribute to falls, and previous research suggests that falls are a significant concern in persons with neuromuscular disease. Wiles and colleagues, for example, studied the prevalence of stumbles and falls in persons with DM1 and found that these patients were 10 times more likely to fall than a control group of people without MD.[19] Similarly, Horlings and colleagues studied falls in persons with FSHD and found that falls were more prevalent in the FSHD group compared to healthy controls.[11] Those who reported more frequent falls were found to have greater muscle weakness than infrequent fallers, to be more unstable when climbing stairs, rising from a chair and standing still with their eyes closed and to have poorer balance control. Furthermore, the falls in the FSHD population were reported from intrinsic causes (patient related), rather than external (environmental) factors, such as an unstable walkway. These findings further support our results on imbalance, and suggest that additional research focus on the relationship between imbalance, weakness and falls prevalence in this population.

Findings from a number of studies indicate that pain is a significant problem for many persons with DM1 and FSHD. [11] Consistent with these observations, we found that pain was indeed present in the majority of our sample, and that it was severe for 24%. Although the majority of participants who endorsed pain reported a worsening of pain since onset, pain was also reported as more likely to remain the same over the previous six month period. This suggests that though pain is more likely to worsen over time, its rate of progression may be slow. Our correlation analyses demonstrated that pain was negatively and significantly associated with our measure of psychological functioning.

The presence of pain in FSHD and DM1 patients as part of the symptom burden is not surprising, yet remains poorly studied. Prior studies indicate that pain and depression can substantially impact social integration and employment rates, and might be as important as physical abilities with respect to these outcomes. A large percentage of FSHD and DM1 patients exhibit elevated scores for bodily pain and depression on standardized testing, including the SF 36, Brief Pain Inventory, and Minnesota Multiphasic Personality Inventory (MMPI) test.[20,21] Indicators of emotional pathology appeared to be associated with chronic pain, whereas physical dysfunction may not be.[21]

Less common in our sample were memory impairment and vision loss. When adjusting for sex, age and duration of MD, only self-reported memory loss was significantly predictive of productive activity, and only in our FSHD sample. This finding is surprising, given that

cognitive difficulties are most commonly associated with DM1 and not with FSHD, and suggests the need for more research exploring associations between patient functioning and measures of cognition and memory in this population. The regression analyses suggest that perceived memory loss is associated with functioning, particularly with productive activities (such as employment). Screening for problems with memory loss in the healthcare continuum might be an effective strategy for moderating the effects of these symptoms on daily life and improve outcomes for patients.

Study Limitations

A number of limitations of the current study should be considered when interpreting the results. First, all of the data was collected as self-report, and this may have introduced some bias and errors, including misinterpretation of questions or symptoms. Future research should use objective measures of symptom severity (e.g., a vision test to assess vision loss) when possible. The questionnaire also asked participants to rate the improvement of their symptoms over the last 6 months and since onset of the symptom. Reporting from past history is not as accurate as assessing the present symptoms and could potentially introduce response error.

The cross-sectional design of the study also makes it difficult to draw causal conclusions regarding the impact of symptoms severity on psychological and physical functioning. Although the lack of significant associations can be used as evidence that a symptom does not have a causal impact on patient functioning, the presence of a significant association does not prove that it does; significant associations are necessary but not sufficient conditions to conclude that a causal relationship exists. The real strength of correlational studies, such as this one, is that they can tell us which factors are more (strong and significant associations) or less (weak and non-significant associations) likely to play a causal role. Thus, they can help determine which factors to target in true experiments that could then be used to learn about causal relationships. In the current study, we identified fatigue, memory loss, and imbalance as the most important symptoms to target in clinical trials and experimental research to determine their actual impact on patient functioning.

As we stated in the Introduction, weakness is the hallmark symptom of MD, and we therefore anticipated that all or virtually all of the study participants would report significant weakness, so did not assess this symptom in the study. Instead, we assessed five additional symptoms that we hypothesized might be important to functioning in individuals with DM1, FSHD, or both. However, in hindsight we now recognize that it would have been useful to also assess the severity of weakness in the sample in order to determine how the symptoms we did assess compare to this hallmark symptom. Future research in this area would do well to assess weakness as well as other symptoms not studied, which could potentially be even more prevalent and impactful than those examined here. These might include anxiety, sleep disturbance, and numbness, among others There may also be other domains of functioning, such as participation or social roles, that may be further impacted by the studied symptoms.

Importantly, a number of factors limit the potential generalizability of study findings. This study only included participants with DM1 and FSHD, just two of many neuromuscular diseases. Although these are the most common in this age demographic, future researchers

should consider examining symptoms in other MD diagnostic groups as well, such as limb girdle, distal and Emery-Dreifuss muscular dystrophy. In addition, the large majority of our sample was white/Caucasian and this may impact the cultural generalizability of our findings. Last, a significant portion of our participants attended the same clinic at the University of Washington. Thus, individuals from a single geographic region were over-represented in the sample. It would be important to replicate this study in other centers to determine the extent to which our findings generalize to the population of individuals with MD.

CONCLUSIONS

Despite the study's limitations, the findings (1) support previous research that pain and fatigue are common in persons with MD; and (2) indicate that imbalance is another common symptom in this population and requires more research. Research is needed to examine in more detail the symptoms that are not as commonly studied, such as imbalance, as well as to study these symptoms in persons with other types of MD. Additional research should also study the efficacy of psychosocial interventions that may improve quality of life and reduce disease burden in this population.

ABBREVIATIONS

MD	Muscular Dystrophy
FSHD	Facioscapulohumeral Dystrophy
DM1	Myotonic Dystrophy Type I
CIQ	Community Integration Questionnaire
NRS	Numerical Rating Scale

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Table 1

Prevalence and Severity of Symptoms: Percentage of participants to rate their symptoms as greater than or equal to one, and greater than or equal to 6 on the 0 – 10 Numerical Rating Scale (0 = none; 10 = very severe).

Frequency of symptoms			
Symptoms	**Frequency (%≥1)**	**Frequency of severe (%≥6)**	**Mean Severity**
Pain	76.5	23.5	3.45 ± 2.74
Fatigue	90.6	46.5	5.14 ± 2.81
Imbalance	82.4	50	4.95 ± 3.25
Memory loss	48.2	3.5	1.49 ± 1.89
Vision loss	43.2	11.8	1.71 ± 2.54

Table 2

Course of Five Symptoms: Percentage of participants indicating the course of their symptoms to worsen, improve, or stay the same in the last six months and since onset of the symptom.

Symptom	Course Since Onset (%)			Course Last 6 Months (%)		
	Worse	Better	Same	Worse	Better	Same
Pain	45.3	6.5	26.5	29.4	3.5	44.1
Fatigue	57.6	2.9	31.2	37.6	1.2	52.4
Imbalance	60.6	1.8	20	37.6	1.8	42.9
Memory Loss	25.3	1.2	22.4	15.3	2.4	31.2
Vision Loss	29.4	4.1	11.2	18.2	4.1	22.4

Table 3

Correlation Coefficients

Symptom	Home Competancy	Social Integration	Productive Activity	Psychologic Functioning
Pain	−0.025	−0.023	−0.101	−0.210**
Fatigue	−0.189*	−0.182*	−0.181*	−0.324***
Imbalance	−0.195*	−0.061	−0.210**	−0.121
Memory loss	−0.189*	−0.245**	−0.258**	−0.225**
Vision loss	−0.194*	−0.268***	−0.236**	−0.207**

* $p < .05$

** $p < .01$

*** $p < .001$

Table 4

Linear regression predicting CIQ social integration, home competency, and SF-36 Mental Health Scales from 5 symptom severity ratings for DM1 and FSHD

Criterion: CIQ Social Integration	***β***	R^2	$R^2\Delta$	$F(R^2\Delta)$
Block 1- Demographic variables		0.044	0.027	2.550
Age	−0.205*			
Sex	0.076			
Duration of NMD	0.089			
Block 2		0.146	−0.102	2.42**
Pain	0.053			
Fatigue	−0.116			
Imbalance	−0.121			
Memory Loss	−0.147			
Vision Loss	−0.152			

Criterion: SF-36 Mental Health	***β***	R^2	$R^2\Delta$	$F(R^2\Delta)$
Block 1- Demographic variables		0.146	−0.007	0.62
Age	0.045			
Sex	−0.052			
Duration of NMD	0.065			
Block 2		0.181	−0.170	3.12***
Pain	−0.099			
Fatigue	−0.354***			
Imbalance	0.023			
Memory Loss	−0.033			
Vision Loss	−0.092			

Criterion: Home Competency	***β***	R^2	$R^2\Delta$	$F(R^2\Delta)$
Block 1- Demographic variables		0.143	0.126	8.66***
Age	−0.083			
Sex	0.325***			
Duration of NMD	−0.131			
Block 2		0.248	−0.105	4.40***
Pain	−0.093			
Fatigue	−0.017			
Imbalance	−0.083			
Memory Loss	−0.103			
Vision Loss	−0.153			

* $p < .05$

** $p < .01$

*** $p < .001$

Table 5

Linear regression predicting productive activity in persons with DM1

Criterion: Productive Activity	β	R^2	$R^2\Delta$	$F(R^2\Delta)$
Block 1- Demographic variables		0.369	0.344	14.83***
Age	−0.247*			
Sex	−0.176			
Duration of NMD	−0.474***			
Block 2		0.274	−0.095	5.35***
Pain	0.023			
Fatigue	0.077			
Imbalance	−0.228			
Memory Loss	0.036			
Vision Loss	−0.267			

* $p < .05$

** $p < .01$

*** $p < .001$

Table 6

Linear regression predicting productive activity for persons with FSHD

Criterion: Productive Activity	β	R^2	$R^2\Delta$	$F(R^2\Delta)$
Block 1- Demographic variables		0.246	0.219	9.22^{***}
Age	-0.425^{***}			
Sex	0.042			
Duration of NMD	−0.175			
Block 2		0.403	0.317	4.66^{***}
Pain	0.047			
Fatigue	0.060			
Imbalance	0.191			
Memory Loss	-0.300^{**}			
Vision Loss	0.088			

* $p < .05$

** $p < .01$

*** $p < .001$