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Patient Identified Disease Burden in Facioscapulohumeral Muscular Dystrophy

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Abstract

Introduction—The multitude of symptoms associated with facioscapulohumeral muscular dystrophy (FSHD) disease burden are of varying importance. The extent of these symptoms and their cumulative effect on the FSHD population is unknown.

Methods—We conducted interviews with adult FSHD patients to identify which symptoms have the greatest effect on their lives. Each interview was recorded, transcribed, coded, and analyzed using a qualitative framework technique, triangulation, and 3-investigator consensus approach.

Results—1375 quotes were obtained through 20 patient interviews. 251 symptoms of importance were identified representing 14 themes of FSHD disease burden. Symptoms associated with mobility impairment, activity limitation, and social role limitation were most frequently mentioned by participants.

Conclusions—There are multiple themes and symptoms, some previously under-recognized, that play a key role in FSHD disease burden.

Keywords

Facioscapulohumeral muscular dystrophy; quality of life; disease burden; neuromuscular disorders; symptoms

Introduction

Facioscapulohumeral muscular dystrophy (FSHD) is an autosomal dominant disorder characterized by bifacial, scapulohumeral, truncal, and lower extremity weakness with relative sparing of the deltoid and forearm muscles.^{1,2} FSHD is the third most common muscular dystrophy with an estimated prevalence between 1:15,000 and 1:20,000.^{3,4} In addition to muscular weakness, individuals with FSHD may have pain associated with

impaired joint stabilization or nerve stretch, hearing loss,⁵ retinal vascular abnormalities (Coat syndrome),⁶⁻⁸ and atrial arrhythmias.^{9,10}

Here we report findings from a series of open-ended FSHD interviews which identify and quantify the most critical issues and symptoms to FSHD patients.

Materials and Methods

We selected subjects over the age of 21 with either genetically confirmed FSHD or symptoms consistent with the diagnosis of FSHD with an affected and genetically confirmed first degree family member. A purposive sampling strategy was utilized to capture a wide range of age and disability.

We conducted in-depth interviews with subjects by telephone. Participants were asked to identify which symptoms of FSHD have the greatest impact on their quality of life (QOL) and describe how their lives are affected by having the disease. All interviews were audio-recorded and transcribed verbatim for later analysis.

Following transcription, 3 investigators independently analyzed the interviews using a framework technique for qualitative analysis.^{11,12} Each interview quote was familiarized, indexed, charted, mapped, matched and interpreted by a team consensus approach.^{11,12} Similar quotes across participants were recorded, and the quote frequency for each symptom was determined. Like symptoms were classified into symptomatic themes of FSHD health, and each theme was further categorized as a physical, mental, social, or disease-specific aspect of FSHD health. Lastly, a model and frequency table was created based on cumulative participant data to highlight the most critical areas of patient-identified FSHD health.

The entire study was conducted with informed consent and institutional review board approval.

Results

Twenty individuals participated in this study. The age of onset, deletion size, education level and occupational status was available for 13 members of our sample (supplemental table 1).

From the 20 FSHD participants, 1375 direct quotes were coded identifying 251 symptoms of importance. These symptoms represented 14 symptomatic themes and 4 major domains (figure 1). The most frequently identified symptoms were “impaired walking” followed by “difficulty with stairs”, “reliance on family members”, “difficulty lifting objects”, “limitations physically on what one can do”, and “difficulty getting places without handicap accessibility”(supplemental table 2). “Difficulty with mobility and ambulation” and “FSHD-specific activity impairment” were the symptomatic themes that had the highest number of representative quotes (figure 1). A full list of participant-identified symptoms, quote frequency, and theme classification is provided (supplemental table 2).

Discussion

This study uses extensive patient interviews to identify the symptoms of highest importance to a sample of FSHD patients. In many instances, these identified symptoms are underreported in the FSHD literature and represent potential areas of therapeutic intervention.

Participants identified poor mobility as a theme that has a major impact on their lives. Advances in orthotic devices as well as improved accessibility to buildings may play a substantial role in further relieving future FSHD disease burden.

The effect FSHD has on a patient's social role and emotional status is often overlooked in the clinical setting, yet these issues have a paramount importance to patients as highlighted through our interviews. Indeed, the frequency that patients mentioned the importance of social role limitations was greater than the frequency of quotes involving the cardinal features of FSHD (fascioscapulohumeral weakness). This is in agreement with other studies that have highlighted the need to address social and emotional factors in muscle diseases and those that have noted a relationship between emotional distress and the perception of disease severity.^{13–15} The emotional and social burdens of FSHD patients should not be neglected, as these aspects may be treatable through social services, social networking programs, education, anti-depressants, psychotherapy, and counseling.

The results from our study supplements prior work evaluating QOL in FSHD.^{13–24} Previous studies have largely used generic instruments (such as the SF-36 and the CIS-fatigue subscale) to characterize the impact of FSHD on patient QOL.^{13–24} Unfortunately, generic instruments may not be suitable for some neuromuscular diseases, as they can exclude questions that are highly relevant to a neuromuscular population while including questions that have limited relevance.¹⁴ While prior studies primarily utilize generic QOL measures to identify a global reduction in overall QOL, increased pain, and worsening fatigue in FSHD, our study further qualifies and quantifies these findings using patients' own words and perspectives.

Previous work has also created and tested generic neuromuscular instruments by combining patient input from diverse neuromuscular populations.^{25,26} The Individualized Neuromuscular QOL measure was initially created using combined interview data from no fewer than 8 distinct neuromuscular populations.²⁵ In our study all interview data came exclusively from FSHD patients. Not surprisingly, there is both overlap and uniqueness when comparing data between studies. For instance, our sample of FSHD patients did not identify "locking" or myotonia as an issue of importance, whereas a combined neuromuscular population including myotonic patients did.²⁵ In addition, compared to myotonic dystrophy type-1 (DM1), FSHD participants report greater difficulty with proximal arm weakness, truncal weakness, facial weakness, and impaired body image.²⁷ Our FSHD participants also identified 113 unique issues not reported by DM1 patients through similar interviews.²⁷ These findings highlight some of the differences between neuromuscular populations and the potential need for disease-specific QOL outcome measures.^{28–31}

This qualitative study was limited by its sample size. Future studies will need to validate the issues identified here using a larger and more diverse FSHD patient sample. All interviews were conducted by telephone. Due to this constraint, strength and functional testing were not performed. Future research will need to determine the association between patient reported disease burden and functional measures as well as age, gender, duration of symptoms, and genetics.

In conclusion, FSHD participants identify a wide array of symptoms that impact their QOL. These symptoms help to define the disease from the patient's perspective and are potentially amendable to future therapeutic intervention.

Supplementary Material

Refer to Web version on PubMed Central for supplementary material.

Acknowledgments

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Abbreviations

FSHD	Facioscapulohumeral muscular dystrophy
QOL	Quality of life
DM1	myotonic dystrophy type-1

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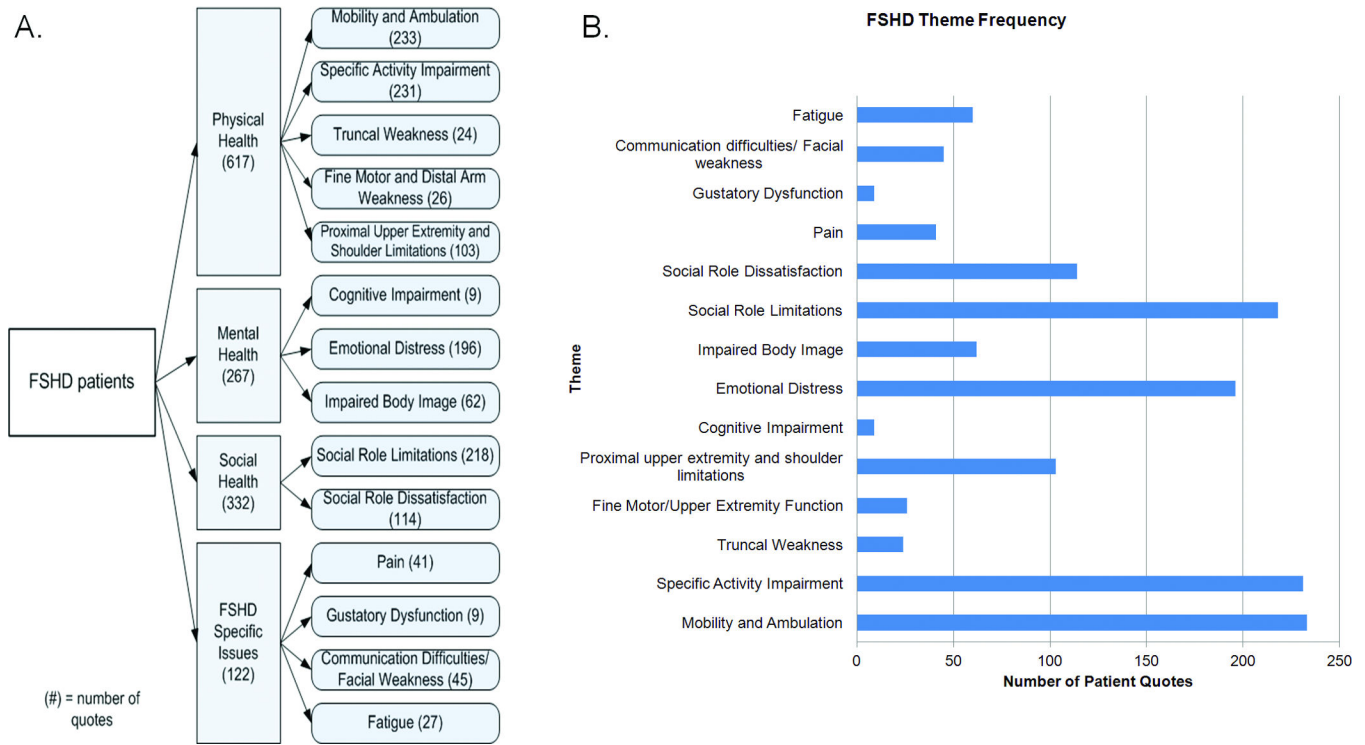


Figure 1. A. Conceptual Model of Quality of Life in FSHD patients as represented by domains, subdomains and themes. Quote frequencies are provided in parentheses. B. Quote frequencies subdivided by subdomain.