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INVITED REVIEW

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The lived experience of facioscapulohumeral muscular dystrophy: A systematic review and synthesis of the qualitative literature

Roisin Murray DClinPsychol[1] | Clare M. Donnelly DClinPsychol[2] | Kent D. Drescher PhD[3] | Christopher D. Graham PhD, DClinPsychol[1,4]

[1]Psychology Service for Older People, Western Health and Social Care Trust, Londonderry, UK

[2]Psychological Therapies Service, Northern Health and Social Care Trust, Antrim, UK

[3]National Center for PTSD, VA Palo Alto Healthcare System, Palo Alto, California, USA

[4]School of Psychological Sciences and Health, University of Strathclyde, Glasgow, UK

Correspondence
Christopher D. Graham, School of Psychology and Health Sciences, University of Strathclyde, Graham Hills Building, Glasgow, Scotland G1 1QE, UK.
Email: christopher.graham@strath.ac.uk

Abstract

Introduction: In this review we sought to characterize the lived experience of people living with FSHD (pwFSHD) to help clinicians to orient their services to the needs of these individuals.

Methods: Five electronic databases were systematically searched for qualitative research studies containing quotations from pwFSHD. ENhancing Transparency in REporting the Synthesis of Qualitative research and Preferred Reporting Items for Systematic Reviews and Meta-Analyses guidelines informed the methodology. Study quality was assessed using the Critical Appraisal Skills Programme Checklist tool, which measures the methodological quality of qualitative research. Data extracted from included studies were analyzed using thematic synthesis.

Results: Ninety-nine pwFSHD took part in the six studies included in this review - from research teams based in two countries. Five descriptive themes emerged: "Engaging with life as symptoms progress"; "The emotional journey"; "A family burden to bear"; "Social connection and disconnection"; and "Tension between visibility and invisibility." From these, two analytical themes were derived: "The emotional challenge of continuing and intensifying adaptation" and "The relational burden of rare disease."

Discussion: The lived experience of pwFSHD is characterized by physical, emotional, and social challenges that impact on engagement with life, particularly as symptoms progress. Further research is needed to provide a fuller understanding of the experience of pain in FSHD and of the lived experience of FSHD across cultures.

Abbreviations: CASP, Critical Appraisal Skills Program Checklist; CERQual, Confidence in the Evidence from Reviews of Qualitative research; ENTREQ, ENhancing Transparency in REporting the Synthesis of Qualitative research; FSHD, facioscapulohumeral muscular dystrophy; PRISMA, Preferred Reporting Items for Systematic Reviews and Meta-Analyses; pwFSHD, people with facioscapulohumeral muscular dystrophy; QoL, quality of life.

The objectives of this activity are to enable the reader to: 1) Understand and be able to apply in patient care and research the biopsychosocial model of quality of life in people with facioscapulohumeral muscular dystrophy and other muscle disorders; 2) Incorporate an understanding of the 5 descriptive themes when caring for individuals with facioscapulohumeral muscular dystrophy; 3) Understand and be able to incorporate into patient care the two analytical themes: a) the emotional challenging of continuing and intensifying adaptation, and b) the relational burden of rare diseases.

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KEYWORDS
facioscapulohumeral muscular dystrophy, lived experience, mood, qualitative synthesis, quality of life, systematic review

1 | INTRODUCTION

Facioscapulohumeral muscular dystrophy (FSHD) is a progressive neuromuscular disease, which is estimated to affect 1 in 15 000 to 20 000 people worldwide.[1] It is characterized by weakness and atrophy primarily affecting the muscles of the face, shoulders, and arms,[2,3] with the lower extremities affected in the later stages of disease progression.[1,4] Common extramuscular symptoms include fatigue and pain.[1,3–5] Onset of symptoms most commonly occurs in the second decade of life. However, there are significant individual differences in disease expression in FSHD, with wide variability in age of onset, rate of progression, symptom severity, and characteristics.[1–3,5,6]

Recommended best practice for the management of FSHD focuses on symptom management to improve functioning and to limit the impact of FSHD on quality of life (QoL).[1,3,6,7] This includes genetic testing and regular neurology review alongside supportive care options such as physical therapy to maximize muscle function and/or reduce pain, and occupational therapy for provision of orthoses, aids, and adaptations for activities of daily living.[6,7]

1.1 | QoL in FSHD

To advance curative treatments and symptom management, research in FSHD has largely focused on understanding the genetic and clinical characteristics of the disease. A body of research has also focused on understanding QoL in FSHD.[8] Research including samples of people with FSHD suggests that, on average, QoL is reduced when compared with those living without the condition. However, there is much variation in QoL between individuals with FSHD.[8] Only a proportion of this variation can be attributed to disease severity,[8,9] and some with very high disease severity have excellent QoL and vice versa. One systematic review of quantitative observational studies showed that factors like mood, pain, and fatigue are consistently correlated with QoL.[8] Thus, it is now understood that a wide range of biopsychosocial factors contribute to QoL.[10] In any given case, QoL may result from a complex interaction of biological factors (e.g., disease severity[9,11]), levels of pain and fatigue,[12] social factors (e.g., employment status[13,14]), and psychological factors (e.g., perceptions,[9] coping mechanisms,[15] values,[16] and mood[17,18]). One implication is that, alongside the aforementioned symptomatic treatments, psychological and social interventions offer additional means for reducing the impact of FSHD on QoL.[10] Indeed, evidence from randomized controlled trials has shown that psychological therapies, such as acceptance and commitment therapy[19] and cognitive-behavioral therapy,[20] can be effective in improving QoL in FSHD.

1.2 | Qualitative research in FSHD

Most research into QoL in FSHD has used quantitative methodologies. This has enabled the benchmarking of QoL in FSHD and an awareness of the breadth of biopsychosocial factors that affect QoL. However, this research provides limited information on how the biopsychosocial factors that affect QoL in FSHD are represented in people's day-to-day lives.

Qualitative research methodologies typically involve interviewing participants from a group of interest, then looking for patterns of recurring themes across accounts. These methods enable an understanding of the experiences of a population of interest. In FSHD, qualitative approaches may therefore enable an understanding of the day-to-day experience of living with this condition. This can provide a more nuanced and granular awareness of the challenges presented by FSHD and how biopsychosocial factors interact to impact QoL. Such information may help health- and social-care professionals to better understand their patients' lives, and tailor their approaches and interventions to more carefully and effectively meet their needs. Therefore, in this review we used established methodologies for synthesizing qualitative literature to detail the experiences of pwFSHD. The specific research question we set out to answer was: *What is the lived experience of individuals with FSHD, in their own words*?

2 | METHODS

2.1 | Design

We undertook a qualitative synthesis employing thematic analysis methodology. We used the ENhancing Transparency in REporting the Synthesis of Qualitative research (ENTREQ) guideline,[21] which presents criteria designed to help researchers accurately describe and report the design, methods, results, and analysis of qualitative data. We also followed the recommendations of the Preferred Reporting Items for Systematic Reviews and Meta-Analyses (PRISMA)[22] statements, a widely adopted guideline that includes criteria designed to help reviewers report the rationale, methods, and results of their reviews with transparency. The review was preregistered on Prospero (ID: CRD42022304258).

2.2 | Search strategy

This systematic review included qualitative and mixed methods studies capturing the lived experience of FSHD. Five electronic databases were searched on February 24, 2022, namely CINAHL, Web of Science, PubMed, PsycINFO, and MEDLINE. The reference sections of

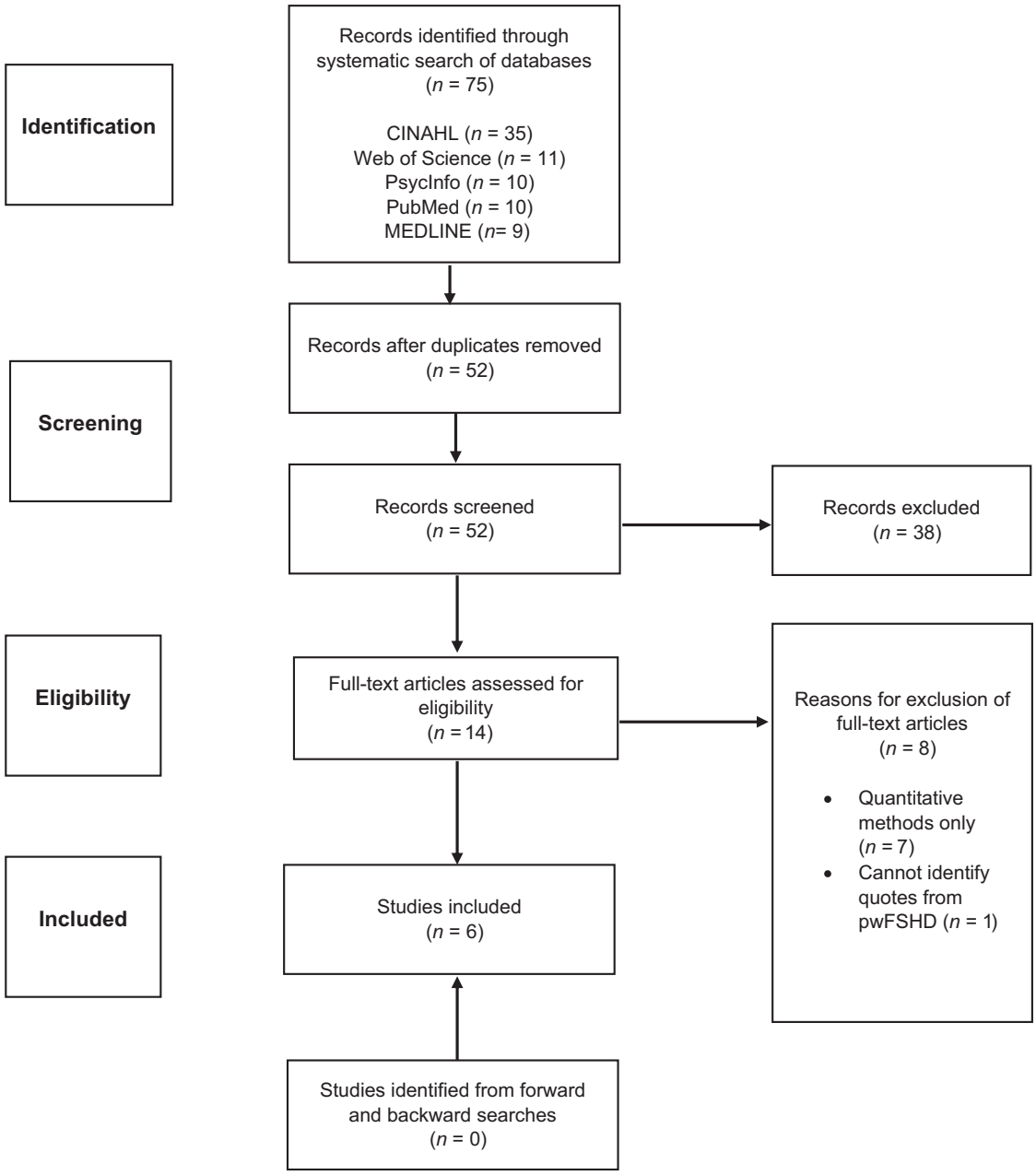

FIGURE 1 Preferred Reporting Items for Systematic Reviews and Meta-Analyses (PRISMA) flowchart.

all included studies were then cross-referenced on Google Scholar using a forward (a search of all studies that have cited an included study) and backward (a search within the reference sections of each included study) strategy.[23] Search terms, informed by the SPIDER tool,[24] were: "facioscapulohumeral muscular dystrophy" OR "FSHD" OR "FSHMD" OR "muscular dystroph*" OR "muscle disorder*" OR "neuromuscular disease" OR "neuromuscular disorder" OR "facioscapulohumeral dystrophy" AND "interview*" OR "focus group*" OR "experience" OR "understand*" OR "attitude*" OR "feel*" OR "view*" OR "opinion*" OR "perce*" OR "belie*" OR "qualitative" OR "mixed method*."

Studies returned by this search strategy were then screened against the following inclusion and exclusion criteria:

Inclusion criteria:

1. Use of a qualitative data collection technique (e.g., interview, focus group, surveys with open questions, observations).
2. Primary research studies, published in a peer-reviewed journal.

Exclusion criteria:

1. Studies that include quantitative data only.
2. Studies in which quotations cannot be directly attributed to people living with FSHD.
3. Studies focusing on the experience of children with FSHD.
4. Review articles.

TABLE 1 Summary of included studies

Study	Country	Aim	Participants	*N*	Age range (years)	Gender	Recruitment, data collection, and analysis methods
Abel et al.[30]	The Netherlands	To obtain insight into the perspectives and experiences of patients, relatives, and clinicians on the outcomes, as well as on the effects and risks of bariatric surgery for people with FSHD and myotonic dystrophy.	Bariatric surgery patients with NMDs, their partner or relative, bariatric surgeons and general practitioners	14 (3 FSHD)	39–47	2F, 1M	Purposive sampling via patient newsletter and direct approach to patients at neuromuscular center. Semistructured interviews Thematic analysis.
Bakker et al.[31]	The Netherlands	To provide better insight in the illness experiences of people with FSHD.	Adults with FSHD	25	24–77	11F, 14M	Purposive sampling via hospitals and rehabilitation centers. Semistructured interviews. Thematic analysis.
Minis et al.[32]	The Netherlands	To explore how persons with NMD still at work perceive the impact of the slow process of NMD-related deterioration on employment participation and the strategies used in maintaining their jobs.	Currently employed adults (aged 18–65 years) with common NMDs.	16 (5 FSHD)	30–62	1F, 4M	Purposive sampling via advertisement within Dutch Federation of Muscular Diseases, directly via neuromuscular center, and by word of mouth. Semistructured interviews. Constant comparison analysis.
Schipper et al.[33]	The Netherlands	To describe how fatigue affects the lives of people with FSHD, how they experience fatigue, and how they deal with it.	Patients with FSHD and severe fatigue	25	24–77	11F, 14M	Purposive sampling from participants in a clinical trial aimed at reducing fatigue and improving QOL for people with FSHD. Semistructured interviews. Content analysis
Sezer et al.[34]	The Netherlands	To gain insight into the functional and psychosocial consequences of facial weakness of patients with FSHD and how they are managing in daily life.	Patients with FSHD and any degree of facial weakness	16	24–77	9F, 7M	Purposive sampling through an announcement in the digital newsletter of the FSHD working group of the patient associations for neuromuscular disorders in The Netherlands and Belgium and directly via neuromuscular center. Semistructured interviews. Grounded Theory.
Yadlin-Segal[35]	Israel/ international	To address the scarcity of research on selfies and disability as a whole, and on selfies in the context of disability and affect in particular.	People with FSHD who participated in the FSHD selfies campaign	25	Not reported	Not reported	Opportunistic sampling. Open-ended questionnaire circulated within FSHD members groups on social media. Thematic analysis.

Abbreviations: FSHD, facioscapulohumeral muscular dystrophy; NMD, neuromuscular disease; QOL, quality of life.

2.3 | Study selection process

The PRISMA flowchart (Figure 1) demonstrates the study selection process. The title, keywords, and abstract of all studies retrieved from searches were uploaded to Rayyan and reviewed by the lead author (R.M.) in terms of the inclusion/exclusion criteria. To ensure reliability and validity in study selection, the second reviewer (C.M.D.) also reviewed 20% of the studies retrieved from searches at random ($n = 11$). Studies that evidently met exclusion criteria from review of the title, keywords, and abstract were excluded. Full texts were then screened. Any articles that were considered for inclusion were independently screened against inclusion and exclusion criteria by the lead author and second reviewer before the final decision was made.

2.4 | Quality assessment

The Critical Appraisal Skills Program Checklist (CASP)[25] is a 10-item tool used to evaluate the quality of studies used in qualitative syntheses and was applied to the studies included in this review. A summary of these assessments is displayed in Table 1. To improve reliability and validity of CASP assessment, two of the studies were assessed independently by two of the researchers (R.M. and C.M.D.) who then met to confirm consensus on quality assessment. The researchers agreed completely on quality assessment ratings. R.M. then independently assessed the remaining four studies. There is variation in how CASP ratings are used to inform analyses within a qualitative synthesis. As is common practice, we used ratings to help us understand the methodological limitations and strengths of primary studies and to contribute to the confidence assessment undertaken as part of the analysis (following GRADE CERQual guidelines[26]) instead of using ratings to exclude studies.[27] The benefit is that the widest body of evidence can contribute to theme development, with quality assessment scores used to weigh relative contributions of each study to analytic themes (confidence assessment).

2.5 | Data extraction

Data extracted from each included study was author(s), year of study, country of study, study aims, methodology and analysis, epistemology, characteristics of the included sample, findings/results, quotations, themes, sampling approaches, and ethical standards. Study results/findings, quotations, and/or themes were extracted verbatim for inclusion in data analysis. As with quality assessment, the first and second reviewers (R.M. and C.M.D.) independently extracted data for a third of included studies ($n = 2$) to improve reliability and validity. A standard data extraction form was developed to guide this process. The reviewers then met to compare and discuss any discrepancies in data extraction. R.M. extracted data from the remaining studies independently ($n = 4$).

2.6 | Data synthesis

Data from included studies was entered into NVivo software and analyzed using thematic synthesis as outlined by Thomas and Harden.[28] We chose to use thematic synthesis because it allows for the synthesis of data across diverse research methodologies and presents results in way that is accessible to health-care professionals.[29]

Stage 1 of thematic synthesis involved coding extracted data line by line. Descriptive themes were then inductively developed by grouping the individual codes together into wider themes. The experiences most frequently and/or emphatically expressed by participants across studies informed the descriptive themes, but consideration was given to individual variability, allowing for the inclusion of differing opinions and experiences. Finally, inductive analytical themes were devised that built on the descriptive themes and more directly answered the research question. The research team, which included R.M. (lead author), C.D.G. (a clinical health psychologist with expertise in FSHD), and K.D.D. (a clinical psychologist who also lives with FSHD), met at two stages to examine the extracted quotations, along with proposed descriptive and analytical themes. Alternative possibilities were explored among the team and the resulting themes refined. Following the GRADE CERQual guidelines,[26] confidence in the analytical themes was assessed in terms of methodological limitations, coherence, relevance, and adequacy.

3 | RESULTS

3.1 | Summary of findings

Six studies were included in this review,[30–35] from research teams based in two countries (Table 1). All studies were published within the last 10 years. In total, 121 people took part in these studies, including 99 people with FSHD. Most involved semistructured interviews,[30–34] and one study collected data via an open-ended questionnaire.[35] Thematic analysis was the most common analysis method,[30,31,35] although one study used constant comparison,[32] one used content analysis,[33] and one grounded theory.[34]

3.2 | Quality assessment

Table 2 shows the results of CASP quality assessment.[25] The average quality of studies included in this review was high. However, two studies[32,34] showed no evidence of "Adequate consideration of researcher–participant relationships" (CASP criterion 6); information on ethical considerations (CASP criterion 7 "Adequate consideration of ethical issues") was limited or unclear in two studies,[34,35] and for one study[33] limited information on data analyses left some uncertainty regarding CASP criterion 8, "Data analysis rigorous."

TABLE 2 CASP quality assessment

Criterion	Studies					
	Abel et al.[30]	Bakker et al.[31]	Minis et al.[32]	Schipper et al.[33]	Sezer et al.[34]	Yadlin-Segal[35]
1. Research aims clear	✓	✓	✓	✓	✓	✓
2. Qualitative methodology appropriate	✓	✓	✓	✓	✓	✓
3. Design appropriate to aims	✓	✓	✓	✓	✓	✓
4. Appropriate recruitment strategy	✓	✓	✓	✓	✓	✓
5. Data collection appropriate	✓	✓	✓	✓	✓	✓
6. Adequate consideration of researcher–participant relationships	✓	✓	X	✓	X	✓
7. Adequate consideration of ethical issues	✓	✓	✓	✓	?	?
8. Data analysis rigorous	✓	✓	✓	?	✓	✓
9. Clear statement of findings	✓	✓	✓	✓	✓	✓
10. Valuable research	✓	✓	✓	✓	✓	✓

Note: ✓ = yes; X = no; ? = cannot tell.

3.3 | Themes

Thematic synthesis revealed five descriptive themes from which two analytical themes were derived. All themes are presented in Table 3.

3.3.1 | Descriptive themes

The five descriptive themes are outlined in what follows. A comprehensive collection of quotations can be found in Table S1.

Engaging with life as symptoms progress

Several common physical symptoms of FSHD were described across studies, including fatigue,[31–33] facial weakness,[34,35] and muscle weakness.[30,31] These symptoms were experienced as having a significant impact on the lives of pwFSHD. However, symptom presentation differed over time and between individuals. Although symptoms may be physically limiting, pwFSHD also described situations in which thoughts or worry about symptoms left them reluctant to engage with different activities.[32]

In response to illness progression, pwFSHD become active problem-solvers, finding strategies to manage the impact of physical symptoms. In the case of fatigue, it was noted in multiple studies that a balance between activity and rest is crucial.[30,32] Although this process of monitoring and adapting to progressive symptoms can be tiring, pwFSHD reported feeling that a good QoL was possible.

The emotional journey

The lived experience of FSHD is not limited to experiencing physical symptoms; a significant emotional impact of illness was also reported across the studies included in this review. This emotional journey begins at diagnosis, where a variety of differing responses were evident, including feeling overwhelmed, relieved, or that the diagnosis offered legitimacy to their symptoms and experience.[30] In contrast, a stage of denial post-diagnosis was reported in several studies.[31,34] This denial speaks to the emotional journey of working toward acceptance of FSHD diagnosis, and may involve feelings of guilt or shame.[30,34]

TABLE 3 Analytical and associated descriptive themes identified through thematic synthesis of six articles

Descriptive themes	Analytical themes
1. Engaging with life as symptoms progress	The emotional challenge of continuing and intensifying adaptation
2. The emotional journey	
3. A family burden to bear	The relational burden of rare disease
4. Social connection and disconnection	
5. Tension between visibility and invisibility	

Following diagnosis, additional challenging emotional experiences were reported including poor self-esteem, low confidence, depression,[29] stress,[32] grief,[34] and anxiety-particularly around the uncertain development and evolution of FSHD symptoms.[30,31,33] In contrast, pwFSHD also sought positive emotional experiences and reported that a positive mental attitude aided coping.[32,33]

A family burden to bear

When FSHD occurs, its repercussions are felt within the family, both in terms of the direct impact of physical limitations, dependency, and care needs,[30,32,33] and the wider impact on family planning, work arrangements, finances, and family relationships.[30,31] The families of pwFSHD must sometimes make sacrifices because of their loved one's condition[30–32] and the changing roles within the family as the illness progresses can be source of stress and emotional distress for pwFSHD.[30–33]

Social connection and disconnection

FSHD can represent both a barrier to and a facilitator of social connection. Although symptoms vary across pwFSHD, it was noted

among several studies that nonverbal communication difficulties associated with facial weakness presented a communication barrier,[30,33,34] particularly when interacting with strangers. Other symptoms such as fatigue may also limit social engagement[32] and employment opportunities.[31] Social isolation was noted as a key issue for pwFSHD,[34] as low prevalence rates create barriers to forming communities with other people who also have this condition. However, a recent social media campaign (#FSHDSelfies) has allowed pwFSHD to connect with an online FSHD community.[34]

Tension between visibility and invisibility

Many pwFSHD do not share the details of their diagnosis with friends, acquaintances, and colleagues,[31,34] and varied experiences of disclosure were outlined across the studies in this review. Both positive and negative experiences were described, ranging from acceptance and support to stigma, bias, and discrimination, highlighting the risk involved in disclosing diagnosis.[31,34] Although some of the symptoms and related adaptations associated with FSHD may be visible, it remains a rare and relatively unknown condition in the general population.[30,34] Moreover, this societal invisibility is mirrored in interactions with health-care professionals, where pwFSHD have encountered a lack of awareness and sometimes have to explain their condition to doctors.[29] Participants in the #FSHDSelfies campaign made several suggestions for improving awareness of the condition, including empowering individuals to take control of cultural representations of FSHD, and increasing visibility of the condition via the use of social media.[35]

3.3.2 | Analytical themes

Two analytical themes were derived from the descriptive themes. As per ENTREQ guidelines,[21] participant quotations are in bold and author interpretations in italic. The first analytical theme, "The emotional challenge of continuing and intensifying adaptation," concerns the ongoing experience of responding to the emerging challenges of living with FSHD and encompasses the two descriptive themes of engaging with life as illness progresses and the emotional journey. The second analytical theme, "The relational burden of rare disease," captures the three descriptive themes of "A family burden to bear," "Social connection and disconnection," and "Tension between visibility and invisibility." This theme embodies the relational experiences that occur in FSHD, which can make it more difficult for pwFSHD to interact with others and participate in a society that is ill-suited to rare disease, while acknowledging the opportunity to connect with a global FSHD community. Table 4 presents a summary of analytical themes and confidence assessments based on GRADE CERQual guidelines.[26]

The emotional challenge of continuing and intensifying adaptation

The impact of FSHD on the individual is far-reaching and ever-changing: **"I can't do the things I would like to do. It is like having a Pac-Man in your life; he's eating and eating till nothing is left"** (p. 1843).[33] As illness progresses, pwFSHD are challenged to adapt to physical changes, their world threatens to become smaller, affecting their ability to engage with life in the way they wish. A diverse and complex range of emotional reactions to symptom changes were described, including guilt,[31] shame,[35] poor self-confidence, depression,[30] stress, and worry.[33] Many of these emotions appeared also to be a response to the uncertain context where it is unknown when and how symptoms will emerge and progress.[31,33,34] To cope with physical symptoms, the uncertainty and the emotions that occur in this context, pwFSHD described a dynamic fluctuation between the use of avoidance/denial methods and acceptance methods. This was first apparent after diagnosis[32,35] and seemed to repeat at stages of illness progression. PwFSHD *"postpone the use of assistive devices for as long as possible"* (p. 982),[31] or find the decision to stop working *"very difficult"* and *"a great loss"* (p. 982).[31] In time, a turning point of acceptance is reached when the physical deterioration can no longer be ignored, "*Many quotes began with the phrase:* ***'I had to be realistic…,'*** *or* ***'I came to realize'*** *indicating being forced by their physical conditions to admit that going on in the old way was no longer an option"* (p. 59).[32]

The relational burden of rare disease

FSHD presents a complex challenge to interpersonal relationships. The impact of FSHD symptoms on communication and speech varies -facial weakness may impact speech for some, with others are less affected.[34] However, nonverbal communication may be affected more significantly. Indeed, participants in one study *"unanimously mention having an aversion to regularly being judged or wrongly understood"* (p. 4)[34] in their interactions with others. PwFSHD may find interacting with others *"tiring"* (p. 4),[34] as they work harder to communicate effectively, avoid misunderstanding, and connect with others. Some pwFSHD report that **"you have no energy for your family, hobbies, or social network"** (p. 59),[32] when managing FSHD. As the illness progresses, pwFSHD may become increasingly dependent on their partners, which *"sometimes put pressure on their relationship"* (p. 982),[31] although many participants *"felt they were able to handle the relationship with their partner quite well"* (p. 982).[31] Illness progression may also lead to a reduction in social interactions, as *"the intensity of social contacts was lowered"* (p. 59),[32] and some pwFSHD were forced to *"make adjustments in their work, others had to change their jobs, and some had to give up work"* (p. 982).[31] With fewer opportunities to connect with others, pwFSHD may become isolated from others and from shared communities.

The experience of invisibility of FSHD can represent further barriers to social connections. Participants described experiencing a societal lack of awareness about FSHD.[30–32,34,35] Thus, the responsibility for education and advocacy falls to pwFSHD who often have to explain their illness across different contexts, including interactions with peers,[33] employers,[32] and medical professionals.[30,32] One pwFSHD remarked **"There are so many FSHers who are unseen for various reasons"** (p. 43).[35]

Previous negative experiences, including discrimination and stigma, may engender a reluctance to disclose an FSHD diagnosis to others. Some pwFSHD have had the experience that *"openness about the disease did not guarantee empathy"* (p. 61)[32] and *"disclosure did not always make things better"* (p. 65),[32] and felt ignored or poorly treated by others,[31,34,35] leaving them feeling more disconnected and isolated.

TABLE 4 CERQual summary of qualitative findings table

Summary of review finding	Studies contributing to the review finding	CERQual assessment of confidence in the evidence	Explanation of CERQual assessment	
1. The emotional challenge of continuing and intensifying adaptation: as FSHD illness progresses, pwFSHD find their engagement with life decreases. Internally, they may experience negative emotions, a dichotomy of denial and acceptance, positivity, being realistic, and an overarching theme of uncertainty. Life with FSHD is a life of constant adaptation.	30–35	Moderate confidence	Methodological limitations Coherence Adequacy Relevance	Moderate concerns No/very minor concerns Minor concerns Minor concerns
2. The relational burden of rare disease: FSHD impacts on relationships with others at the individual, group, community, and societal levels. The nature of invisibility of rare disease is an major barrier to overcome through education and awareness, a burden that falls to pwFSHD.	23–28,30–35	Moderate confidence	Methodological limitations Coherence Adequacy Relevance	Moderate concerns No/very minor concerns Minor concerns Minor concerns

Abbreviations: FSHD, facioscapulohumeral muscular dystrophy; pwFSHD, people with FSHD.

4 | DISCUSSION

Five descriptive themes cover the breadth of biopsychosocial factors that have been implicated in quantitative studies of QoL[5,8]: "Engaging with life as symptoms progress"; "An emotional journey"; "A family burden to bear"; "Social connection and disconnection"; and "Tension between visibility and invisibility." However, these themes add nuance to our understanding of QoL by showing how the different variables interact to impact QoL. For example, the analytical theme, "The emotional challenge of continuing and intensifying adaptation," demonstrates the interaction between symptom progression and psychological factors. In response to emerging symptoms, participants described having a range of emotional reactions. Alongside this the uncertainty regarding the rate and severity of symptom progression is a context that naturally promotes anxiety. In coping with changing symptoms and the associated emotional reactions, participants describe oscillating between the use of denial and acceptance strategies. These methods of coping can have varying consequences on QoL. For example, denial of symptoms and emotions may be helpful initially for controlling difficult emotions (reducing anxiety, embarrassment, etc.). However, denial-based coping may also cause reduced awareness of changes in mobility and the necessity to adapt one's environment to maximize functioning. As mobility worsens, denial-based coping methods may therefore lead to limited functioning and poorer QoL. Participants communicated that they eventually reached a point where they needed to accept physical changes to find methods for maximizing functioning (e.g., use of mobility aides). This suggests that acceptance strategies for symptoms and associated emotions are eventually used by pwFSHD as alternatives to denial strategies and that this facilitates better functioning.

The processes identified in this theme closely cohere with the biopsychosocial model of QoL in muscle disorders that we suggested some years ago (Figure 2),[10] as specified in Diagram B of Figure 2. This demonstrates that muscle disorder symptoms challenge individuals to find instrumental and emotional coping responses, with acceptance versus denial the central coping responses for bettering or worsening QoL. In support of the centrality of acceptance coping methods for QoL, a quantitative observational study showed this was significantly associated with life satisfaction and mood in muscle disorders,[16] and a recent randomized controlled trial of a psychological intervention that promotes acceptance-based strategies showed efficacy for improving QoL and mood in muscle disorders.[19]

The other analytical theme, "The relational burden of rare disease," demonstrates how FSHD symptoms can challenge the quality of relationships. This can have an impact on longstanding intimate relationships, where changes in mobility can lead to requirements for support. It can also have an impact on forming new relationships, as facial weakness/visible physical difference leads to concerns about communicating effectively with others and the ability to form friendships and intimate relationships. There is also an additional communication burden placed on pwFSHD: FSHD is a rare disease, which means that the symptoms involved, treatment options, and key personal challenges are not widely understood by society. Thus, in interactions with others, even health-care professionals, pwFSHD often need to educate them about FSHD and their corresponding needs. This analytical theme demonstrates how a fuller range of biopsychosocial

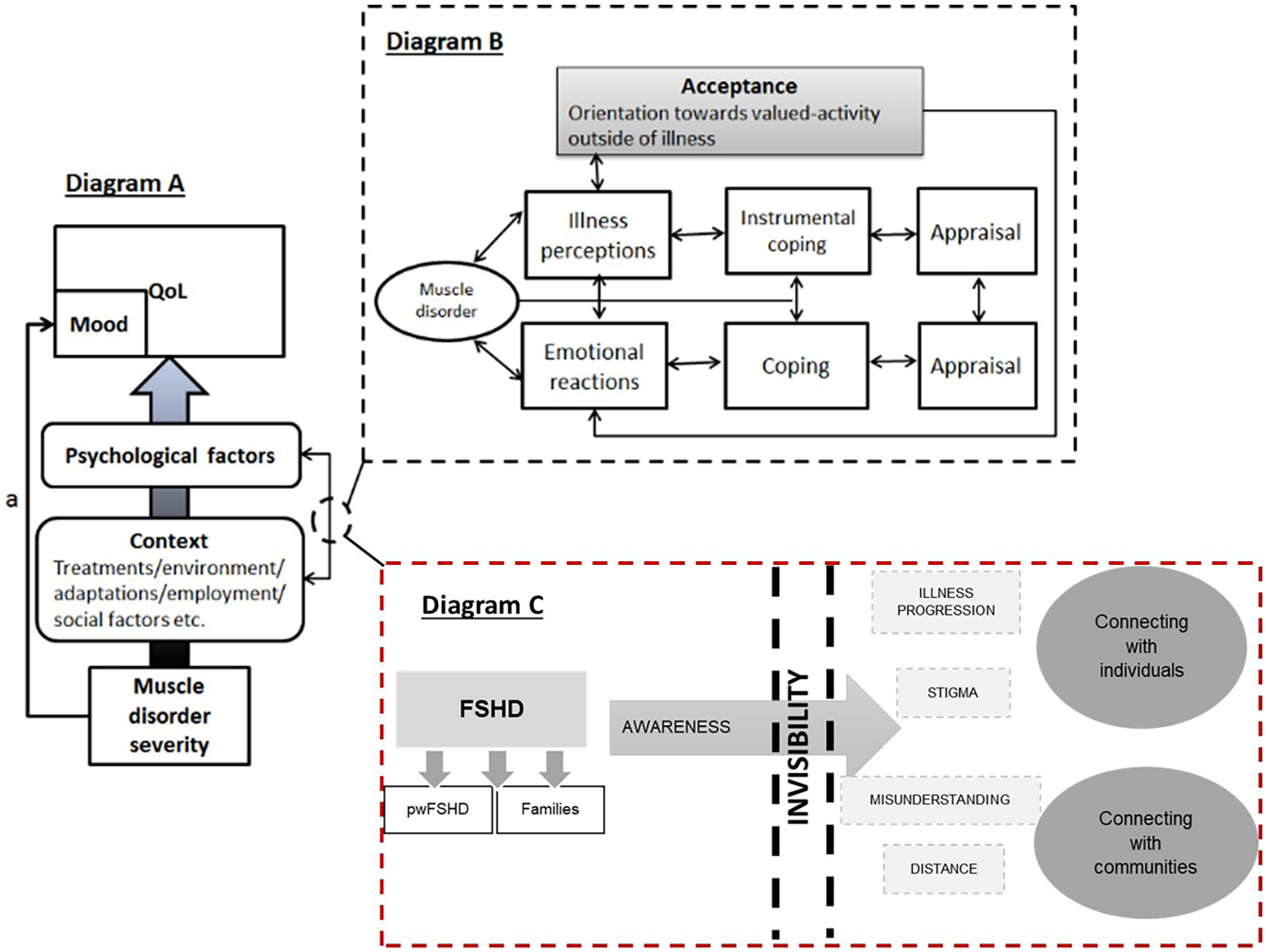

FIGURE 2 The biopsychosocial model of quality of life (QoL) in muscle disorders, extended to include the analytic theme "The relational burden of rare disease" (Diagram C). Diagram A: Although some biological factors may directly impact on QoL and mood (e.g., autonomic dysfunction and central nervous involvement, path a), in general, the impact of muscle disorder severity is mediated by psychosocial variables. Diagram B: Focusing on psychological factors. An adapted version of Leventhal's self-regulatory model, showing how an individual's perceptions of their illness (e.g., the symptoms involved and the potential for the illness to be controlled with treatment or behavior) influences the instrumental coping methods they use to manage the symptoms. There is a parallel process of managing emotional responses to illness, which includes finding ways to accept uncomfortable emotions in order to undertake meaningful activity. Diagram C: Focusing on social factors. PwFSHD can share the burden of illness with their families. The disease may become a barrier to connecting with others on an individual and community level. Although raising awareness can break through this barrier of invisibility, other obstacles, such as stigma, lack of understanding, illness progression, and physical distance, can continue to affect relationships for pwFSHD.

factors interact to impact on QoL. Changing symptoms (biological) challenge the quality of relationships (social), which, understandably, is accompanied by normal emotional responses that stimulate coping methods (psychological). A qualitative review of illness experiences in other rare diseases also noted a range of themes showing how a condition can have a complex impact on relationships,[36] such as themes detailing tensions regarding how and when to disclose a condition to others, a sense of sameness and difference to others, and experiences of stigma. This particular aspect of the lived experience of FSHD has been less thoroughly researched in quantitative studies of QoL. However, a large, comprehensive study by Johnston and colleagues that aimed to collate the key functional impacts of FSHD[5] showed that 75% of participants endorsed the idea that FSHD leads to concerns about reliance on family. The centrality of this experience among people with FSHD in the qualitative literature suggests that the biopsychosocial model of QoL should be updated to emphasize relationships as a central process (Diagram C in Figure 2 shows the updated model).

4.1 | Clinical implications

Exploration of acceptance and denial-based coping methods is a key part of a psychological intervention shown to be effective for improving QoL in FSHD.[19] In addition, an oscillation between denial and acceptance coping appears relevant to struggles around the use of mobility aids, and thus could also be explored by occupational

therapists, physiotherapists, and others when discussing changes in mobility and options for enhancing functioning.

The theme, "The relational burden of rare disease," suggests that interventions targeting connection and reducing isolation may also be of benefit to pwFSHD. Promoting links with an international online FSHD community could provide an influential social intervention to reduce isolation, improve social connection, and build a peer support network for pwFSHD. An example of such an approach is the #FSHDselfies campaign, which represented an important community-building exercise for pwFSHD and also facilitated acceptance of identity as an "FSHer."[34] Further, it appears that a wider awareness of FSHD, and what this condition involves, would reduce some of the burden of explaining the condition to others, including health-care professionals. Thus, programs that facilitate a wider public awareness of FSHD are indicated.

4.2 | Limitations

Comparing our findings with those of the Johnson et al. study,[5] which aimed to establish the full range of functional impacts of FSHD, highlights that some important experiences of pwFSHD may have been missed within existing qualitative research. First, pain is a common symptom, identified by a large proportion (87.7%) of pwFSHD,[4] and evidence shows that pain has wide-reaching effects on well-being and functioning.[8,12] However, experiences of pain were notably absent from the six studies included in this review. Likewise, a significant proportion of participants in the study by Johnson et al.[5] identified issues of cognitive impairment such as deficits in memory (30% of participants) or concentration (10%), phenomena not noted within the studies in this review. The absence of these key themes from the existing qualitative research emphasizes the need for additional research exploring the experience of pwFSHD.

A notable limitation of this systematic review is the small number of qualitative studies available for thematic synthesis, some of which discussed very specific aspects of FSHD and therefore offered limited insight into the research question. However, both analytical themes were derived from data extracted from all six included studies and confidence in these findings based on CERQual framework[26] was deemed to be moderate (see Table 4). The breakdown of male-to-female participants was reasonably balanced, and the age ranges included adults in their middle 20s to late 70s. However, future research should occur across a wider range of countries–to evaluate the extent to which findings generalize. The present qualitative synthesis provides a frame of reference for future research to consider similarities and differences in the illness experience of pwFSHD across cultures.

The Biopsychosocial Model of QoL in FSHD (Figure 2), particularly the newly proposed addition presented in Diagram C, should be considered exploratory and future research is required to examine the applicability of the model. The review was interested in the perspectives/lived experience of pwFSHD, and did not examine the views of family members or relatives. Future research of the lived experiences of significant others of pwFSHD would provide a fuller understanding of the wider impact of FSHD beyond the individual with FSHD.

4.3 | Conclusions

To describe the experience of living with FSHD, this systematic review synthesized data from 99 pwFSHD, who participated in six studies. PwFSHD experience constant physical and emotional adaption to an uncertain illness progression, typified by an "emotional challenge of continuing and intensifying adaptation" as FSHD threatens engagement with life. FSHD also brings a "relational burden of rare disease," with direct and indirect barriers to communication with others, ranging from an inability to smile to being geographically separate from other people experiencing the same condition, and a lack of awareness regarding FSHD Future work is required to better understand the lived experience of pain in FSHD and the experiences of FSHD across cultures.

AUTHOR CONTRIBUTIONS

Roisin Murray: Conceptualization; data curation; formal analysis; investigation; methodology; project administration; resources; software; validation; visualization; writing – original draft; writing – review and editing. **Clare M Donnelly:** Formal analysis; methodology; validation; writing – review and editing. **Kent Drescher:** Conceptualization; formal analysis; investigation; methodology; supervision; validation; writing – review and editing. **Christopher Graham:** Conceptualization; formal analysis; investigation; methodology; resources; supervision; validation; visualization; writing – original draft; writing – review and editing.

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As a systematic review, all data sources are identified and can be collated by others.

ETHICAL PUBLICATION STATEMENT

We confirm that we have read the Journal's position on issues involved in ethical publication and affirm that this report is consistent with those guidelines.

ORCID

Christopher D. Graham https://orcid.org/0000-0001-8456-9154

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SUPPORTING INFORMATION

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

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