European Journal of Paediatric Neurology 50 (2024) 64–73

Contents lists available at ScienceDirect

European Journal of Paediatric Neurology

journal homepage: www.journals.elsevier.com/european-journal-of-paediatric-neurology

Original article

Quality of life and support needs in children, adolescents, and young adults with facioscapulohumeral dystrophy, a mixed-method study

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ARTICLE INFO

Keywords:
Facioscapulohumeral dystrophy
Quality of life
Neuromuscular disease
Children
Adolescents
Mixed-method study

ABSTRACT

Background and objectives: Quality of life (QoL) in children with facioscapulohumeral dystrophy (FSHD) seems plausible decreased. Little is known about factors influencing QoL in children with FSHD. Our objective is to explore factors contributing to the QoL of children, adolescents, and young adults with FSHD, to describe how they experience life with FSHD, and to report their support needs.
Methods: We performed a mixed-method study with individual age-appropriate semi-structured interviews assessing QoL in children, adolescents, and young adults with FSHD and their parents. To characterize the sample, quantitative data on QoL, pain, fatigue, and participation were collected. Interview data was analyzed using a thematic analysis.
Results: Fourteen patients participated (age between 9 and 26 years old, eight males and six females). The degree of FSHD severity, as indicated by the FSHD-score, did not correlate with QoL. Older children had a lower QoL than younger children. Children and adolescents strived for normality regardless of physical discomfort. Phenotypical features of FSHD led to insecurity aggravated by hurtful comments of others. The unpredictability of disease progression and its implications for career and parenthood choices led to a generalized feeling of uncertainty about the future. Support was found within family and friends. Participants expressed a need for peer support and psychological support as well as recommending it to others.
Discussion: Quality of life in childhood FSHD is diminished caused by their physical limitations, altered appearance, fear of social rejection, and uncertainty of the disease progression in the future. A fear of social rejection most likely contributes to striving for normality regardless of physical discomfort. Support should be focused on acceptance and coping with hurtful comments. It should preferably be individualized, easily accessible and not offered as therapy but rather as tutoring for children.

1. Introduction

Facioscapulohumeral dystrophy (FSHD) is one of the most prevalent hereditary autosomal dominant neuromuscular diseases and is caused by a repeat contraction of D4Z4 on chromosome 4q35 [1]. In approximately 20% of the FSHD patients, symptom onset starts in childhood [2, 3]. Childhood FSHD is characterized by facial and scapular weakness, pain, and fatigue in most children, however, the severity varies widely. The prevalence of childhood FSHD is estimated at 1 in 100,000 in the Netherlands [4].

Our recent prospective study found a reduced quality of life (QoL) in 70% of children with FSHD [4], measured by the Kidscreen-52

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https://doi.org/10.1016/j.ejpn.2024.04.006
Received 12 October 2023; Received in revised form 26 January 2024; Accepted 15 April 2024
Available online 21 April 2024

questionnaire [5]. The QoL in FSHD was more reduced than in boys with Duchenne muscular dystrophy, even though the physical disease severity is generally lower in FSHD [4,6]. Several hypotheses have been suggested to explain this [4,7]. First, childhood FSHD used to be perceived as synonymous to the severely affected early-onset FSHD subgroup. However, our study found a milder spectrum of FSHD phenotype resembling the clinical spectrum of classic FSHD [4]. The relatively invisible phenotype of childhood FSHD might be less often recognized as a muscle disease and thus results in higher expectations. Furthermore, facial weakness might influence social contacts and communication, which is a known burden in other diseases [8,9]. Moreover, qualitative research on the influence of facial weakness in adults showed that facial weakness significantly affects many aspects of daily life [10]. However, research seeking to understand the determinants of QoL in children and adolescents is lacking.

A decreased QoL in childhood may have a substantial effect on children's development, school performance and social life. Additionally, the disease severity gradually progresses over time, which may further reduce QoL over time. Finally, adult FSHD patients reported lack of support in childhood and advised psychosocial support for younger patients [10,11].

This study therefore aims to explore (1) which factors contribute to the QoL of children, adolescents, and young adults with FSHD; (2) how they experience life with FSHD; and (3) to obtain insight into their support needs. We expect that our findings provide insight in the psychosocial support needs of the children, adolescents, and young adults, which may guide the development of future intervention programs.

2. Methods

A mixed-method study was performed using semi-structured interviews with children, adolescents, and young adults diagnosed with FSHD and their parents to get insight in their QoL. The clinical disease severity was assessed during regular hospital visits with the FSHD clinical score (range 0–15, with higher scores indicating more severe disease) [12]. The study was approved by the Medical Ethics Committee of the Radboud University Medical Centre (registration number: 2022–13794). All participants and parents signed a written consent form and provided consent for the use of previously collected questionnaire data from the iFocus research [4]. The qualitative part of this research including the design and execution followed the Consolidated Criteria for Reporting Qualitative Research (COREQ) checklist [13].

2.1. Recruitment and study population

Between July 2022 and October 2022, children, adolescents, and young adults (age 6–26 years) with genetically confirmed FSHD were approached for participation. Subjects with insufficient command of the Dutch language were excluded. No additional exclusion criteria existed. Participants were recruited by purposive sampling and assessed for eligibility from the iFocus cohort (nationwide natural history study of children with FSHD) [4,7] and from the (child)neurology outpatient clinic of the FSHD National Referral Centre. All eligible patients were invited to participate. Consent for review of medical files (D4Z4 repeat length; FSHD clinical score) was obtained.

2.2. Data collection

Participants and their parents were asked to fill in questionnaires on: QoL, pain, fatigue, and participation (data and method triangulation to ensure credibility).

The Kidscreen-52 was used to assess the QoL in children between 8 and 18 years of age. This questionnaire was constructed, tested, and normed using data from 3000 healthy children and adolescents from Europe. This automatically enabled comparison with data of healthy children and adolescents. Parents were asked to complete the proxy version of the Kidscreen-52 to assess the agreement on QoL between children and their parents. The Kidscreen-52 questionnaire consists of ten subscales: Physical wellbeing, Psychological wellbeing, Moods & Emotions, Self-perception, Autonomy, Parent relations & Home life, Financial resources, Social support & Peers and School and Social acceptance. The questions are rated by a five-point Likert scale with higher scores indicating a higher QoL [5]. Scores are displayed as a z-score (number of standard deviations from the mean).

The 36-Item Short-Form Health Survey questionnaire version 2 (SF-36v2) was used to assess QoL in adolescents of 18 years and above and consists of eight domains: Physical functioning, Role limitations due to physical health, Role limitations due to emotional problems, Vitality, Mental health, Social functioning, Bodily pain and General health. The questions are rated by a five-point Likert scale with higher scores indicating a higher QoL and scores are presented as z-scores [14].

The NeuroQoL fatigue domain in a child and adult version was used to assess fatigue and consists of eight questions rated by a five-point Likert scale with higher score indicating more fatigue [15].

The Pediatric pain questionnaire was used to quantify pain for children below the age of 18 and the McGill was used for adults. Both questionnaires are composed of 35 different words to describe pain and

Table 1
Questionnaires used per scored item for children, adults and parents.

	<18 years of age	>18 years of age	Parents
Item	Questionnaires		
Quality of life	Kidscreen-52[11] *(10 domains)*	SF-36v2 [12] *(eight domains)*	Kidscreen-52 proxy version
Pain[b] (VAS 0–10)	Pediatric pain [14] questionnaire	McGill pain [15] questionnaire	
Fatigue	NeuroQoL fatigue [13] children	NeuroQoL fatigue adults [13]	
Participation	USER-P children [16] *(three scales)*	USER-P adults [16] *(three scales)*	

Legend.
Kidscreen-52 questionnaire consists of ten subscales: Physical wellbeing, Psychological wellbeing, Moods & Emotions, Self-perception, Autonomy, Parent relations & Home life, Financial resources, Social support & Peers and School and Social acceptance. The questions are rated by a five-point Likert scale with higher scores indicating a higher QoL [5].
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The NeuroQoL fatigue domain in a child and adult version was used to assess fatigue and consists of eight questions rated by a five-point Likert scale with higher score indicating more fatigue. [15].
The Pediatric pain questionnaire was used to quantify pain for children below the age of 18 and the McGill was used for adults. Both questionnaires are composed of 35 different words to describe pain and a visual analogue scale (VAS) to rate pain between 0 and 10.[16,17].
The USER-Participation in child and adult version was used to assess participation three scales: frequency, restrictions and satisfaction. Scores per scale are converted to a scale ranging from 0 to 100, with higher scores indicating better levels of participation meaning higher frequency, less restrictions and higher satisfaction. [18].

a visual analogue scale (VAS) to rate pain between 0 and 10 [16,17].

The USER-Participation in child and adult version was used to assess participation on three scales: frequency, restrictions, and satisfaction. Scores per scale are converted to a scale ranging from 0 to 100, with higher scores indicating better levels of participation meaning higher frequency, less restrictions and higher satisfaction [18].

(Table 1 and Supplementary data C).

Qualitative data was collected via semi-structured in-depth interviews (via videoconference). Parents were invited to participate in the interview of participants of 6–11 years. For participants aged 12–17, children choose whether to have a parent present during the interview. A literature- and expert-based age-appropriate interview guide with open-ended questions was composed in close collaboration with the FSHD patient organization (AL, AR) (Supplementary data A). All interviews were conducted by the same researcher (prolonged engagement to ensure credibility) (HB) who was unrelated to any of the participants. After the first interview, the interviewer (HB) and research team (JD, NR, EC, SA) discussed the data to refine the interview skills of the interviewer and adapt the interview guides to improve the quality of the subsequent interviews (audit trail). Data collection continued until data saturation was reached after 6 weeks and 14 interviews [19]. The researcher kept field notes during and after the interviews (to ensure transferability and reflexivity). Data collection and analysis was an iterative process to improve validity. Interviews were transcribed verbatim and anonymized (HB). Video recordings were eventually destroyed. After two weeks an interview summary was sent to the participant as a member's check for feedback.

2.3. Data analysis

Quantitative data was analyzed using SPSS version 22 (IBM SPSS Inc, Chicago IL). Data was assessed for normality assumptions using the Shapiro-Wilk test. Continuous parametric variables (questionnaires) were expressed as standard deviations from the mean as z-score using established reference values [20]. The Wilcoxon signed rank test was used to measure agreement on QoL between children and their parents. The Mann-Whitney *U* test was used to compare Kidscreen-52 data of this cohort to a non-participating group of nine children with FSHD, which participated earlier in the iFocus 2-year follow-up study. Correlations between QoL, FSHD severity and age were analyzed using Spearman's rho. A *p*-value $<0,05$ was considered statistically significant.

The anonymized transcripts were analyzed using Atlas.ti version 22. Data analysis was thematic [21]. The data analysis was inductive and consisted of three phases: 1) transcripts were read and reread and open coding was used to familiarize with the data and to label common concepts amongst the interviews with codes closely related to the participants' words. 2) Two researchers (HB, SA) independently analyzed and coded the interviews (investigator triangulation to ensure credibility). Codes were discussed among the two researchers and consensus was reached to accomplish intersubjectivity. Axial coding was used to identify relations between the open codes and group those into categories (code book in Supplementary data B). 3) Relations and patterns between categories were investigated and described during selective coding. Themes and subthemes were discussed with the research team (consisting of (child) neurologists (in training) (NV, CE, JD, NR), nurse practitioner (SA), master student (HB), and FSHD patients (AL, AR)).

3. Results

In total, 28 patients were approached for the interviews, 14 of whom did not want to participate due to various reasons (Fig. 1). The mean age and disease severity of these patients did not differ from the included participants. Fourteen children, adolescents, and young adults were included (age range 9–26 years (median 17 years); eight males, six females). All participants were ambulatory (Table 2).

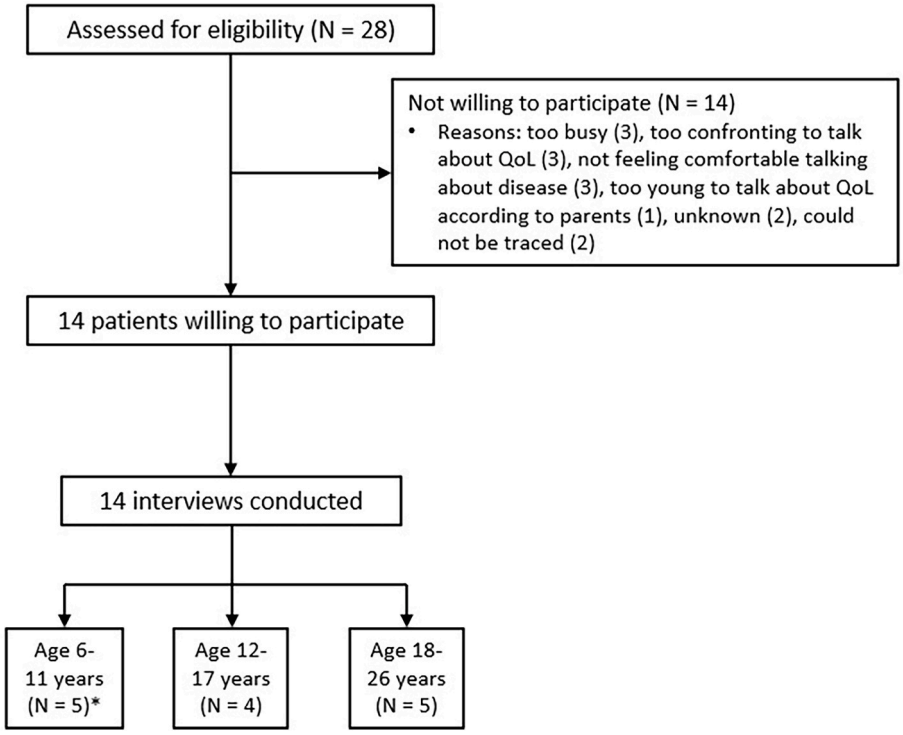

Fig. 1. Flow diagram of inclusions
Fig. 1: * One child did not complete the questionnaires but participated in the interview.

Table 2
Overview of participant characteristics, in ascending order of age.

Participant number	Sex/ Age	D4Z4 repeats	FSHD clinical score	Parent present during interview	Current health status	Social setting	School/work/ other
1	M, 9	5	3	Yes	Mild facial weakness, bodily pain during exercise	Lives with parents, parent with FSHD, one healthy sibling	Special education
2	M, 9	3	9	Yes	Severe facial weakness, reduced shoulder abduction, fatigue	Lives with parents, only person in family with FSHD	Primary school
3	F, 10	8	2	Yes	Fluctuating shoulder pain, mild facial weakness	Lives with parents, parent with FSHD. Six siblings, three with very similar symptoms but were not tested for FSHD	Primary school
4	M, 10	2	5	Yes	Mild leg pain when walking	Lives with parents, parent and sibling with FSHD, one healthy sibling	Primary school
5	F, 11	6	3	Yes	Pain in shoulders, fatigue, mild facial weakness	Lives with parents, parent and grandparent with FSHD	Primary school
6	F, 13	2	4	Yes	Mild facial weakness	Lives with parents, parent and sibling with FSHD, one healthy sibling	Secondary school
7	F, 17	2	7	No	Mild facial weakness, reduced shoulder abduction.	Lives with parents, only person in family with FSHD	Vocational college
8	M, 17	5	6	Yes	Pain in shoulders and legs, fatigue, severe facial weakness.	Lives with parents, parent and grandparent with FSHD, one healthy sibling	Secondary school
9	M, 17	2	6	No	Reduced shoulder abduction, mild facial weakness	Lives with parents, only person in family with FSHD	Secondary school
10	F, 19	4	4	No	Mild facial weakness, occasionally pain in shoulders, fatigue	Lives in dorm, only person in family with FSHD	College
11	M, 20	4	6	No	Mild facial weakness, fatigue, leg and shoulder pain during exercise	Lives with parents, parent with FSHD	College and parttime work
12	F, 22	7	4	No	Reduced shoulder abduction, fatigue	Lives with parents, uncle with FSHD, parent with mild symptoms but was not tested, 2 healthy siblings	Full-time job
13	M, 22	8	3	No	Reduced shoulder abduction and occasionally pain in shoulders	Engaged, lives with partner, parent and one sibling with FSHD, two healthy siblings	Full-time job
14	M, 26	8	10	No	Fatigue, pain in neck and shoulder during activities, reduced shoulder abduction, very mild facial weakness	Lives with partner, only family member with genetically proven FSHD, father and uncles have symptoms but were not tested	Full-time job

3.1. Participants' characteristics

3.1.1. QoL, fatigue, pain, and participation in children, adolescents, and young adults with FSHD

Details of the questionnaire results are shown in Table 3. The mean QoL assessed by the Kidscreen-52 was higher (z-score 0,4) in this cohort compared to healthy children. A lower score was found in the domain Social Support & Peers (z-score −0,3). In adolescents and young adults, the 36-Item Short-Form Health Survey questionnaire version 2 (SF36v2) showed a lower QoL (z-score −0,4) compared to the healthy population. Lower scores were found in the domains: Physical functioning, Role limitation due to physical health, Social functioning, Bodily pain and General Health. Lower overall QoL was observed in the non-participating group of nine children, as evidenced by the comparison of Kidscreen-52 results from the included participants with those who had previously participated in the iFocus 2-year follow-up study (which overall included 20 children with FSHD) and did not want to participate in the current study [4]. Although not statistically significant, they scored lower in the domains: Social Acceptance (mean z-score −0,3), Physical well-being (mean z-score −0,2) and Psychological well-being (mean z-score −0,2).

The NeuroQoL fatigue questionnaire showed that this cohort experienced little fatigue (z-score −0,4). Pain was frequently reported (77%), mainly in the shoulders, upper legs and calves (mean pain score in the past week of 2,5 (range 1–6, SD 2). The USER-P questionnaire showed that participants were overall satisfied with their participation (mean 90%, SD 11) and experience little restrictions (mean 80%, SD 11).

3.1.2. Correlation between QoL, disease severity and age

Disease severity (FSHD clinical score) ranged between 2 and 10 (mean 5 ± 2 SD). No correlation was found between QoL (z-score of Kidscreen-52 and SF36v2) and disease severity ($r = -0{,}12$, $p = 0{,}7$). An older age was correlated with a lower QoL ($r = -0{,}55$, $p=0{,}049$).

3.1.3. Comparison of the subscales of QoL between children and adolescents with FSHD and their parents

The Kidscreen-52 overall showed a good agreement between the scores of children and parents, except for the Social support & peers domain: parents scored their children higher (z-score 0,3), while children scored lower (z-score −0,3) compared to a healthy population. Although not statistically significant, the parents scored lower than their children on the domains: Physical well-being, Moods & Emotions and especially Social acceptance (Table 3).

3.2. Qualitative results

Fourteen interviews were conducted with children (in seven interviews a parent was present), adolescents, and young adults with FSHD (Fig. 1). The duration varied between 37 and 92 min. Six major themes were identified.

3.2.1. Acceptance and what it means to have FSHD

3.2.1.1. FSHD interferes with everyday life. Participants explained that FSHD is part of their everyday life and that many choices depend on their physical restrictions. The extent to which they felt that FSHD has an influence on their daily life, ranged from 'I barely notice it' to 'FSHD makes all the choices'. Some participants mentioned that FSHD was diagnosed when they were very little and has been incorporated in their life and therefore, they felt only few restrictions.

> *"I told my mom that if I could ever make a wish and I could wish that the muscle disease would go away, I wouldn't do that, I would make another wish. […] I would wish that we would become happy."* (Participant 5, age 11, F)

Other participants were more recently diagnosed and felt that they still struggled with acceptance and frustrations every day.

Table 3
QoL measured by Kidscreen-52 and SF36v2.

	N children/adolescents	Mean SD children/adolescents[a]	N parents	Mean SD parents[a]	*P*-value[b]
Kidscreen-52 total (participants < 18 years of age)	8	0,4	8	0,2	0,61
Physical well-being		0,2		−0,2	0,49
Psychological well-being		0,5		0,6	0,31
Moods & emotions		0,1		−0,2	0,86
Self-perception		0,3		0,1	0,49
Autonomy		0,5		0,8	0,49
Parent relations & home life		0,6		0,8	0,61
Financial resources		0,9		0,3	0,93
Social support and peers		−0,3		0,3	**0,04**[c]
School		0,9		0,1	0,06
Social acceptance		0,1		−0,7	0,49
SF36V2 (participants > 18 years of age)	5	−0,4		–	–
Physical functioning		−1.5		–	–
Role physical health		−0,4		–	–
Role emotional problems		0,3		–	–
Vitality		0,2		–	–
Mental health		0,01		–	–
Social functioning		−0,2		–	–
Pain		−0,5		–	–
General health		−1,4		–	–

(FSHD: facioscapulohumeral dystrophy, SF36V2: 36-Item Short-Form Health Survey questionnaire version 2).
[a] Compared to healthy subjects.
[b] Wilcoxon signed-rank test.
[c] Statistically significant.

"I guess I'm frustrated that I'm being held back by something I can't do much about." (Participant 14, age 26, M)

Some children choose different hobbies and sports due to physical limitations. Furthermore, going out with friends or going on holidays required much more planning and participants were not always able to take part in these activities.

"Going out for one day is fine, but the other day my friends and I went to a festival for three days and that was too much, I was sick with fatigue." (Participant 10, age 19, F)

Feelings of sadness and frustration were mentioned when participants talked about the loss of independence due to the progression of the disease. One participant described it as 'a reality check' when the strength in his legs muscle decreased and he was required to take a step back in his work and social life. He explained that once he had accepted the loss of a certain muscle function, the next setback was already around the corner.

"You have barely time to take it all in, you keep losing more function, it goes way too fast and you are way too young for this." (Participant 14, age 26, M)

Participants did not want to ask for help when they were unable to do things because of their physical restrictions, instead they tried to find solutions themselves.

3.2.1.2. Striving for a normal life. Even though participants experienced physical consequences of FSHD daily, they all wanted to be treated like their healthy peers and strived for a normal life. They did not want to be the center of attention or be treated differently.

"I just want my life to be as normal as possible. I would really hate it if I would end up in a wheelchair. I would score my life a straight 3 instead of an 8 now. I really just want to look as normal as possible and be able to do the things everyone else does." (Participant 7, age 17, F)

When participants told others about their disease, they tried to avoid the term 'muscle disease', as they all felt that the meaning of that word is too severe for their condition.

[…] "I really have to explain it, if they really want to know, beause *muscle disease sounds like a severe form of disease, and I really do not like that term."* (Participant 9, age 17, M)

When describing what a normal life would look like, participants used their friends's lifes as a comparison. Some participants would contemplate about what their life would be without FSHD. Others felt that they were being confronted with their disease on a daily basis and considered their physicial limitations as the cause of a decreased QoL.

"If I didn't had this disease, I think I would rate my quality of life much higher." (Participant 11, age 20, M)

3.2.1.3. Dealing with functional restrictions in daily life. Different coping strategies were used. Often participants ignored their physical discomfort and dealt with the consequences of fatigue and pain afterwards. Most participants found it difficult to listen to their body's limits and did not want to be the only one that needed a break during sports or other activities. They felt frustrated about their restrictions since they did not want to be different from their peers and felt they missed out on all the fun stuff. They rather pushed through at the expense of their energy balance.

"It feels like giving up, and that feeling bothers me more than the feeling of being tired." (Participant 14, age 26, M)

Overall, participants tried to approach their physical restrictions as positively as possible and tried to put things in perspective when frustrated. They tend to look for solutions and think in possibilities.

"I often feel better when I put things in perspective and try to make the best of it, but it still bothers me sometimes." (Participant 14, age 26, M)

3.2.2. Appearance and self-image

3.2.2.1. The influence of facial weakness and winged scapulae on appearance and self-image. Concerns regarding physical appearance, stemming from facial weakness and altered scapular features, were predominantly articulated by older females. Almost none of the participants above 10 years of age liked their photo to be taken or liked their appearance on a photo. They described their faces as neutral and weird looking or they were not pleased with their smile.

"I am not happy when I look at my own picture, because I still *have the idea that I have a crooked face. My friends are very sweet and tell me that I'm beautiful the way I am, but for me that is very difficult."* (Participant 10, age 19, F)

Some children therefore avoided pictures and selfies, others were very picky about which photo they would post on social media.

"When my picture is taken, I say: I don't have a good smile, or take another, or I don't look good. […] I just want to know how my real smile looks." (Participant 5, age 11, F)

Furthermore, mainly girls felt insecure about the presence of scapular winging, and did not want others to see them.

"My shoulder blades look different; I always have two points sticking out of my back. That bothers me in the summer when I wear a bikini. I'm afraid that people will see that, so I have grown my hair and let it fall over my shoulder blades. I would never wear a ponytail in the summer, I always have my hair down." (Participant 7, age 17, F)

Some participants do not feel bothered by their facial weakness and feel that their appearance has no influence on their self-image, they accepted FSHD as part of their life.

"I am who I am and my muscle disease is part of that." (Participant 8, age 17, M)

However, in other children, the facial weakness and altered appearances of the shoulder blades has an influence on their self-image.

"When I look in the mirror, I sometimes think: Why do I have this? Why does it have to be me having a muscle disease?" (Participant 6, age 13, F)

3.2.2.2. Negative comments on facial features and expression. Some participants received negative comments about their face. Many of these comments were regarding their facial expression and included comments like*: 'don't look so angry'*, *'you look grumpy'* and *'you should smile more'* other comments were regarding children's lips and involved annoying questions and remarks about the use of Botox and fillers.

"Other children think I used fillers. […]. There were two girls in my class and they told everyone I use Botox." (Participant 6, age 13, F)

For some children, these comments were painful, and they would not understand why people would make these comments.

"Last year, there was a girl in my class, and she said that I look like a fish […] That was very unkind, and I thought, why would she say that?" (Participant 6, age 13, F)

Other children said that they did not care very much about their lack of facial expression and the comments made by others as it was not something they could change, and they didn't feel restricted because of it.

"Now and then I notice that they cannot read the expression from my face, that is difficult sometimes, but you know, that doesn't really hinder me." (Participant 8, age 17, M)

Children and adolescents reported little problems with verbal communication, only some participants mentioned that their voice was sometimes soft or that they would mumble, this did not cause any problems.

3.2.3. FSHD affects social interactions

3.2.3.1. Fear of social rejection. For children below the age of 12, their biggest concern was the fear of social rejection when telling other children and adults (such as teachers and sports trainers) about their muscle disease. They feared that they would be treated differently, would be disqualified from physcial activities or being bullied.

"I am afraid of that, that if other children know, someone will say: you should stay inside, because you have a muscle disease." (Participant 5, age 11, F)

However, only few children described actual experiences of social rejection.

[…] "He is afraid to go to school on his tricycle because he is afraid of being laughed at. If he runs, he is afraid other children will make comments on that." (Father of participant 2, age 9, M)

There were multiple opinions amongst participants and their parents regarding informing others about FSHD. None of the participants would inform others inmediately when meeting new people, although some participants felt comfortable explaining their muscle disease if one was interested. Several participants would rather not tell other people about their diagnosis for various reasons. One participant explained that when he would talk to his friends and collegues about FSHD, he would declare himself 'a patient' and accepting that was very difficult. Some participants did not want to burden classmates or collegues with the diagnosis; they would only disclose in case of implications for specific (work or sport related) activities. A minority of participants only felt comfortable talking about their muscle disease with their close friends.

3.2.3.2. Invisibility causes misunderstanding, prejudice and unrealistic expectations. In many participants, the FSHD phenotype was not visible at a first sight. Most considered this advantageous, especially in striving for a normal life. Other participants felt that it also has a disadvantage, since they often ended up in situations where they were unable to live up to expectations due to their physical limitations.

"There are certainly times, when I have to say: I know it sounds weird but I'm unable to do that. […] But also, for example smaller stuff, like in the supermarket, when an older lady asks me to get something from the top shelf, I have to tell her: no, I can't." (Participant 14, age 26, M)

Mainly parents believed that due to the invisibility and ignorance, judgment by others was common and caused children to stretch their bodies' limits.

3.2.4. A future with FSHD

3.2.4.1. Uncertainty about the future. When asking how they would see their future, almost all participants above the age of 17 talked about the uncertainty of the progression of their muscle disease. Most of them were worried about losing their independence due to deterioration of weakness or ending up in a wheelchair. The diverse phenotype and difference in progression amongst FSHD patients, made it difficult to see a future perspective.

"I am scared for the future; it is so uncertain." (Participant 10, age 19, F)

"On bad days, all I can think of is wheelchairs and very weak faces." (Participant 14, age 26, M)

Others felt that there was no point in worrying because they were not able to change the future and would take is as it comes. Furthermore, participants with currently no or limited progression, were more optimistic about their future. To get some idea about the speed of decline, some participants searched for comparison amongst family members with FSHD.

3.2.4.2. Inheritance for a subsequent generation. An important topic mentioned by the older children and adolescents was the consequences of FSHD for a subsequent generation. Most participants had a positive

view on having children in the future. This was mainly because they experienced only mild inconveniences of the FSHD. They did not consider their QoL less due to the FSHD. Other participants were more ambivalent about a future with children considering their own disease progression as well as possible inheritance of the disease for their future child. Some participants considered preimplantation genetic testing or surrogacy as a solution to prevent inheritance.

> *"Especially the inheritance, that is something you always keep with you, because nobody knows what the future will hold, not for me, but also not for any future children."* (Participant 13, age 22, M)

> *"It will always be a question, because I will become weaker and weaker probably. Am I able to be the father that I want to be? That is a very difficult question. Maybe a question you only get answered by just doing it. But yes, that has to be a well thought-through decision."* (Participant 14, age 26, M)

3.2.4.3. Career options. Many of the children experienced some degree of physical restrictions due the FSHD. They took this into account while making a choice on their future education. Children were aware that they were not able to endure an education or career with many physical aspects. Furthermore, careers with lots of social interaction were also considered difficult. Some participant felt sad that their preferred education or career would not be feasible. Most of them felt comfortable in the education and career they had chosen although it was not their first choice.

> *"I thought about becoming a sign language interpreter, but then I thought, that will become too difficult as my facial muscles will only deteriorate in the future and you are required to make certain facial expressions. […] and then I thought about becoming a primary school teacher, but working in a classroom all day will be too tiring. […] But I am happy with my education now."* (Participant 10, age 19, F)

3.2.5. Support needs of children, adolescents, young adults and their parents

There was a clear difference in opinions on support needs between children with parents who were also diagnosed with FSHD and children whose parents did not have FSHD.

3.2.5.1. Support by family and friends. All participants, regardless of their parents having FSHD, felt that friends and family are most important in sharing their troubles, doubts and worries. A good network of friends was felt important to live a normal life. The support of family was considered valuable to share their experiences and emotions.

> *"Fortunately, I have a lot of good people surrounding me, family and friends that support me very well. I think that you need that, especially in the beginning, to be able to talk about it."* (Participant 10, age 19, F)

In families with more FSHD patients, participants found recognition and answers to questions. Participants from FSHD families did not feel that they needed any extra support.

> *"At the moment, I don't need any extra support. I can find the support I need in my social surroundings and for me that is enough."* (Participant 8, age 17, M)

3.2.5.2. Peer support. Participants who did not have family members with FSHD, expressed a wish for peer support. They explained that it could help them to talk about their disease and be able to learn coping skills from other patients (e.g., on disclosure and career possibilities) as they were not able to find this information within their family. One father expressed his wish for parent-peer support.

> *"When I had a lot of muscle tests to find the diagnosis, I had a friend who also had had these tests. It turned out she didn't have FSHD, but at that time I had an ally, so to speak, we could talk about our experiences together, we understood each other. […] I would like that [peer support], if my symptoms worsen, to learn how others deal with it."* (Participant 10, age 19, F)

3.2.5.3. Psychological support regarding acceptance. A few older participants received psychological support shortly after FSHD was diagnosed and explained that it provided help regarding acceptance and future perspective.

> *"I personally highly recommend that [psychological support], I would have liked that for myself, if I could have had that earlier. […] I would also be interested in having some kind of group sessions maybe, with other adolescents with FSHD. Not right now, because I think I'm not mentally ready for that, but I think that could be very educational and helpful."* (Participant 10, age 19, F)

Many participants did not yet see any added value of psychological support and were confident they were able to deal with their emotions and acceptance themselves.

3.2.5.4. Support by the National Referral Centre. Many participants and parents felt supported by the National Referral Centre. For them it is a place they can reach out to in case of questions or the need for medical support.

> "Without the Nation*al Referral Centre, I wouldn't have any medical guidance, it's just really nice to have that." […] "I'm able to talk with my family and friends for support, but I know I can always reach out to the professionals of the National Referral Centre whenever I need to."* (Participant 13, age 22, M)

3.2.5.5. Need for information about medication trials. Many parents and some older participants mentioned their wish for information about current clinical trials (in children). They were worried that children would be left out. Parents understood that developing medication for FSHD will take time and that it must be approved for adults first. However, they still liked to be updated.

3.2.6. Parental handling

3.2.6.1. What the future will hold. The unpredictability of the future is not only an important topic for the older children and adolescents but even so for their parents. Parents worry about the disease progression and consequences for their child's mobility, future careers and relationships.

> *"How will it go? I don't know. The progression is slightly different for everyone. No one knows how fast his progression will go. That makes it so uncertain."* (Father of participant 1, age 9, M)

Some parents hope that medication will quickly become available for their children. For one father with FSHD it was the reason he participated in the medicine trials for adult FSHD patients. Seeing their children decline caused feelings of sadness and frustration.

> *"In my experience, he is always deteriorating, but I realized that other children are moving forward in their development, and he is standing still."* (Father of participant 2, age 9, M)

Other parents tried not to worry much about their child's future and tried to put things in perspective. They felt confident they would take life as it unfolded and would be able to get the support when they needed it.

3.2.6.2. Protecting their child. When asked upon, parents did not feel that they were more protective over their child compared to other parents or handled their child with FSHD different from their siblings. However, during the conversations with parents who had FSHD, they emphasized that they wanted to avoid issues they had faced themselves in their youth regarding school, work and sports and would therefore take extra measures to be ahead of suspected problems.

> *"At gymnastics in school, I would never get a high grade, because I was unable to do certain* excercises*. I only realised when I was older that it was due to FSHD. I have always said that I want to avoid that for my child. […]. That's the reason I got him tested for FSHD. I didn't want him to run into those things that bothered me. I wanted to prevent that for him."* (Mother of participant 8, age 17, M)

Besides that, during the interviews, parents used verbal and non-verbal communication (out of sight of their child) to express towards the interviewer that certain topics should not be discussed with their children.

4. Discussion

This mixed-method study on QoL in children, adolescents, and young adults with FSHD showed that QoL was lower in the older participants but did not correlate with FSHD clinical severity. The interviews provided insight in how individuals experience life with FSHD, revealing multiple factors that the participants described as influencing their QoL. First, they wanted their lives to be as normal as possible although FSHD affected many aspects of it. Missing out on enjoyable time with friends due to fatigue was considered most bothersome, although often patients participated anyhow and ignored their physical discomfort. Facial weakness and scapular winging were held responsible for insecurities and negative self-image reinforced by upsetting comments by others. The unpredictable disease course led to a feeling of uncertainty about the future (e.g., inheritance and career options). This was concerning for most adolescents and parents. Support was found within family and friends and at the FSHD National Referral Centre. It is expected that peer support will be helpful for some children and adolescents and recommend psychological support for coping with limitations. The main results will be discussed in more detail below.

Striving for normality and fear of social rejection. Qualitative data showed that, just like their healthy friends, children, and adolescents with FSHD strived to live a normal life, consequently ignoring physical discomfort (pain and fatigue). In research on children's experiences living with various long-term conditions, striving for normality regardless of physical discomfort was one of their main findings [22,23]. This might be related to the fact that social acceptance is an important aspect in every child's life to develop social skills regardless of health status [24]. The younger participants described a fear of social rejection when disclosing FSHD, in line with studies in children with other chronic disease [22]. In a qualitative study with chronically ill adolescents, perceived social rejection and the fear of being seen as different was one of the main reasons to withhold disease disclosure. Likewise in this study, immediate family members and close friends were the main group of people to open-up to [25].

Disability paradox. The invisible phenotype of FSHD was both seen as an advantage and disadvantage. The invisibility helped in striving for normality but simultaneously caused unrealistic expectations and situations in which a child was forced to explain its physical limitations or stretch physical limits. The invisibility of the disease may play an important role in the 'disability paradox', a phenomenon describing that patients with severe impairments tend to report higher QoL [26]. Children and adolescents with FSHD may strive for an ideal healthy state which is reinforced by the invisibility of the disease and the fear of social rejection even though this ideal state might not be achievable. This combined with overestimation by their social surroundings seems to have a large impact on their QoL, which was supported by the lower scores in the Social support & Peers and Social functioning domains in the questionnaires.

Insecurities about physical appearance. Facial weakness and winged scapulae were important sources for insecurities about physical appearance in some older children and adolescents (mainly females), which was aggravated by hurtful remarks of other. Physical appearance is important for all children and adolescence transitioning into adulthood, especially since social media is an integrated part of life and shows appearance-focused content [27,28]. The gender difference in experienced insecurities has been previously reported in adult FSHD patients and other facial disfiguring diseases and might be caused by the difference in focus on physical appearance [10,29].

Uncertainty of the disease progression. Uncertainty of the disease progression in the future is a major topic for adolescents and parents. Most children and adolescents think that they would rate their QoL lower in case of future wheelchair dependency. Previous literature has linked illness uncertainty to psychological distress and a decreased QoL in children and adolescents with a chronic and terminal illness [30,31]. Although FSHD is not a terminal disease, constant uncertainty about the future and the possible loss of independence may have a substantial influence on their QoL. A qualitative study in adult muscular dystrophy patients described that constant loss of physical abilities due to disease progression causes stress and influences one's identity [32].

Parental handling. Parents tend to protect children with chronic diseases more than healthy children [33]. Parents trying to manage what and how children are informed about their illness have been described in other studies and can both facilitate and constrain communication with children [34]. By using nonverbal communication during the interviews, parents tried to direct the topics discussed. Furthermore, interviewing children and parents separately revealed notable differences in children's and parents' view on social support and social acceptance. This observation was supported by discrepancies on the Kidscreen-52 questionnaire and previous research [6,35]. It emphasizes the importance of talking with children separately in a clinical setting. It enables clinicians to provide better care and should start early in the transition to adult care.

Support needs. Support was mainly found within family and friends and was considered important to feel accepted and understood by others. Children from FSHD families generally feel confident in their ability to independently handle and address their own problems and challenges. Conversely, adolescents without FSHD family members reported the wish to meet other FSHD patients. Literature showed that higher levels of perceived social support have been associated with better psychological functioning [36]. Support should be tailored to the specific needs of the individual and consist of a focus on modes of acceptance, dealing with disease progression, functional impairment and social interaction. Therapies should be age-specific, easily accessible, and preferably not associated with hospitals or (medical) treatment, as this may lower the bar for children and adolescents to seek support without immediately feeling like 'a patient' or 'ill'.

There is a broad spectrum of clinical severity within muscle diseases ranging from chronic to terminal. FSHD is a rather unknown muscle disease and children showed a lot of ignorance regarding their disease. The lack of knowledge about the variety of clinical severity within muscle diseases may be due to the larger social media campaigns about terminal muscle diseases resulting in connotations with 'fast decline' and 'death' by the public. These assumptions may hinder social participation and integration for patients with a chronic or milder muscle disease. Social media campaigns about muscle disease should also address the importance of social participation to promote social acceptation.

Strengths and limitations. This study had some limitations. First, only part of the cohort was willing to participate in the study and the quantitative data did not show a decreased QoL in children below 18 years. This was in contrast with the findings of our recent cohort study [4,7]. QoL might be a sensitive topic for children and adolescents with a

decreased QoL and therefore they might be less likely to participate in this study. Combined with social desirability bias, this could be an explanation for this Kidscreen-52 results. To test this, we compared the Kidscreen-52 results of the participants with that of children from the iFocus 2-year follow-up study who refrained from participation. This showed a lower overall QoL in the latter group (Results section 3.1). Therefore, selection bias is likely, and the results discussed before might be an overestimation of the actual QoL in children and adolescents with FSHD. Furthermore, the parents' presence in seven interviews may have limited the children to speak freely out of loyalty towards their parents [37]. The online interviews might have been both a strength and a weakness, since it made easier and more accessible to participate but might have led to less detailed information. A strength of this study is the use of the COREQ checklist. For example, the interview guides were compelled together with FSHD patients and clinicians and were based on previous research [4,7]. An international survey on QoL among children and adolescents with FSHD would contribute to the generalizability.

5. Conclusion

This mixed-method study in children, adolescents, and young adults with FSHD showed that FSHD affects different aspects of children's and adolescent's life. They tried to have a positive approach regarding their physical limitations and altered appearance, even though comments of others were upsetting. Striving for normality regardless of physical discomfort might be due to the fear of social rejection. The uncertainty about the disease progression is worrying. Support should be focused on acceptance, dealing with insecurities and hurtful comments, and should be individualized, easily accessible and not framed as 'therapy'.

Declaration of competing interest

Authors have no conflict of interest to declare.

Acknowledgements

The authors like to thank all children, adolescents, young adults, and parents that participated in this study for their time and effort. We would like to thank Prinses Beatrix Spierfonds and Spieren voor Spieren for funding the iFocus research, which resulted in the design and execution of this study. Furthermore, we would like to thank Ietske Siemann for her previous work for the FSHD patient organization and Anouska Ramaker (AR) for her feedback on the interview guides. Several authors of this publication are members of the Netherlands Neuromuscular Center (NL-NMD) and the European Reference Network for rare neuromuscular diseases (EURO-NMD).

Appendix A. Supplementary data

Supplementary data to this article can be found online at https://doi.org/10.1016/j.ejpn.2024.04.006.

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