



**Facioscapulohumeral  
Muscular Dystrophy (FSHD)**

# VOICE OF THE PATIENT REPORT

**Externally Led  
Patient-Focused  
Drug Development  
Meeting**



*This report is dedicated to the individuals  
who courageously shared their stories.*

**AUTHORS:** Jamshid Arjomand, PhD, Chief Science Officer, FSHD Society; Lisa Bain, Medical Writer; June Kinoshita, Director of Research and Patient Engagement, FSHD Society; Mark Stone, President and Chief Executive Officer, FSHD Society

**FUNDING AND ADVOCACY PARTNERS:** Friends of FSH Research, Fulcrum Therapeutics, Muscular Dystrophy Association, Optum

**CONSULTING PARTNERS:** James E. Valentine, JD MHS, and Larry Bauer, RN MA, Hyman, Phelps & McNamara, P.C.

**CLINICAL ADVISOR:** Kathryn Wagner, MD PhD, Kennedy Krieger Institute and Johns Hopkins School of Medicine

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**POINT OF CONTACT:** June Kinoshita, [june.kinoshita@fshdsociety.org](mailto:june.kinoshita@fshdsociety.org)

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## 1. Report summary

Facioscapulohumeral muscular dystrophy (FSHD) is a serious, rare, heterogeneous, progressive, and disabling type of muscular dystrophy. The name reflects the stereotypical muscle groups affected – those of the face (facio), shoulder blades (scapula), and upper arm (humerus), although lower limb and abdominal muscles are also often affected. The resulting muscle weakness may result in mobility impairment, chronic pain, loss of function and independence, and psychological distress. There is currently no cure for FSHD, and no treatments to prevent or stop muscle wasting and weakness. Management thus focuses on relieving symptoms and maintaining function.

In June 2020, the FSHD Society convened a virtual Voice of the Patient Forum to enable the U.S. Food and Drug Administration (FDA) and other important drug development stakeholders to hear directly from patients and caregivers about their journey with FSHD. The meeting was organized as part of the FDA's Externally Led Patient-Focused Drug Development (EL-PFDD) Initiative, which was first established by the FDA and Congress in 2012 to "more systematically obtain the patient perspective on specific diseases and their treatment." The PFDD program was designed to directly hear from patients and families about the major symptoms they experience and the impact of those symptoms on their daily lives, as well as what treatments they are currently using and the unmet needs not addressed by those treatments. This information can aid drug developers and the FDA review staff during the development of new therapies as well as in the review of marketing applications for new drugs.

Prior to the FSHD EL-PFDD meeting, nearly 600 people from all over the world completed a survey about the impact of FSHD on their lives. More than 90% of the respondents were individuals living with FSHD. Parents, guardians, and others who are caregivers or represent adults with FSHD comprised about 8% of respondents. The data from this survey were used to shape the meeting and develop discussion questions designed to collect information helpful to other families living with FSHD, as well as to drug developers and regulators.

The pre-meeting survey data, polling data, remarks from panelists and other live virtual participants, and comments submitted online or by email after the meeting provided the content for this "Voice of the Patient" report. Key themes emerging from these data are summarized below and explored in greater detail in the report.



## Key themes emerging from the survey, meeting, and follow-up comments

### **FSHD is a progressive disease with the potential for severe consequences across the lifespan.**

- The core symptoms of FSHD involve muscles of the face and upper and lower extremities.
- Muscles of the pelvic girdle and abdomen may also be affected.
- Respiratory insufficiency caused by FSHD may shorten the lifespan in some patients.

### **FSHD is highly variable and heterogeneous in presentation.**

- While FSHD typically manifests in adolescence or early adulthood, severe and rapidly progressing early-onset forms may affect infants and young children.
- Impaired mobility, difficulty using hands and arms, fatigue, and pain may significantly reduce physical functioning, quality of life, and the ability to live independently.
- Weakness of the facial muscles resulting in an inability to smile, speak clearly, or fully close the eyelids may not be recognized as symptoms of FSHD until other symptoms appear.
- FSHD is also associated with bilateral sensorineural hearing loss and retinal telangiectasia or Coats' disease.
- The progressive nature of FSHD can contribute to social isolation and loss of employment, and can heighten anxiety and depression.
- Diagnosis of FSHD requires both clinical and genetic assessments.

### **Management of FSHD focuses primarily on symptomatic treatments.**

- There is no FDA-approved treatment for FSHD that prevents or slows muscle wasting and weakness.
- Pharmacological treatments are used to treat pain, depression, and other symptoms of FSHD.
- Exercise, physical therapy, assistive devices, and other non-pharmacological and lifestyle approaches may help patients maintain function as the disease progresses.
- Both pharmacological and non-pharmacological treatment approaches have limited effectiveness, may have substantial side effects, and may be inaccessible to patients due to physical and financial barriers.

### **The lack of treatments and progressive nature of the disease highlight the unmet medical needs in FSHD.**

- FSHD patients are supportive of treatments that halt or slow the progression of the disease.
- FSHD patients welcome restorative treatments that improve muscle strength and function.
- People with FSHD are eager to work with drug developers and regulators to expedite the approval of new safe and effective treatments.



# YOUR VOICE MATTERS



## 2. Introduction

On June 29, 2020, in the shadow of the COVID-19 pandemic, the facioscapulohumeral muscular dystrophy (FSHD) community came together virtually for a Voice of the Patient Forum, organized by the FSHD Society and co-sponsored by the Muscular Dystrophy Association (MDA), Optum, Fulcrum Therapeutics, and Friends of FSH Research. This Externally-Led Patient-Focused Drug Development (EL-PFDD) meeting was designed to allow the FDA to hear directly from patients and caregivers about their journey with FSHD, as well as what would constitute a meaningful therapeutic intervention. Following the model established by the FDA for EL-PFDD meetings, the online meeting welcomed approximately 400 participants, including people with FSHD, caregivers, representatives of the FDA, industry, academia, and other medical product development stakeholders. This day was unique in that it was devoted entirely to listening to and learning from individuals living with FSHD and their caregivers.

Founded by individuals with FSHD in 1991, the FSHD Society has worked to accelerate FSHD research by catalyzing a global community of families, clinicians, researchers from the biopharmaceutical industry and academia, and government agencies such as the FDA to work together toward the goal of developing treatments for this devastating disease. In 2019, the Society convened a landmark meeting to introduce the FDA to the unmet medical needs of people with FSHD and the recent advances in clinical trial readiness and infrastructure.<sup>1</sup>

There has been a long history of successful PFDD meetings in neuromuscular diseases, including Duchenne muscular dystrophy, myotonic dystrophy, Friedreich's ataxia, and spinal muscular atrophy. These meetings have been instrumental in ensuring that the patient voice is reflected in the development of safe and effective drugs for specific disorders.

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<sup>1</sup> *Industry Collaborative for Therapy Development in FSHD*. Published by FSHD Society on June 11, 2019. Available online at [fshdsociety.org/wp-content/uploads/2019/06/FSH-Society-Industry-Collaborative-Report.pdf](https://fshdsociety.org/wp-content/uploads/2019/06/FSH-Society-Industry-Collaborative-Report.pdf).





### 3. Overview of FSHD and its management

FSHD is one of the most common muscular dystrophies, with a prevalence of about one in 8,333<sup>2</sup> to one in 10,000 individuals,<sup>3</sup> or between 33,000 and 40,000 individuals in the United States. Like other muscular dystrophies, FSHD is a genetic disease; however, the genetic mechanisms underlying FSHD are more complicated than in some other muscular dystrophies. Most people with FSHD (about 95%) have Type 1 disease, which is usually inherited in an autosomal dominant manner. This means that in most cases a person has inherited the disease-causing mutation from one parent, and that children of an affected individual have a 50% chance of inheriting the mutation. About 10% to 30% of individuals with FSHD Type 1 have *de novo* mutations, which means they are new and not inherited from a parent. About 5% of people with FSHD have Type 2 disease, which is inherited in a digenic manner, indicating the involvement of two different genes.<sup>4</sup> Although the genetics and molecular mechanisms underlying Type 1 and 2 are different, both result in the abnormal expression of the *DUX4* gene, which leads to muscle cell death and atrophy.<sup>5</sup>

FSHD is progressive and widely variable in severity. It can be extremely disabling. Respiratory insufficiency in FSHD can be life shortening. Symptoms typically appear in adolescence or early adulthood as a skeletal muscle disease. Less frequently, infants and younger children may also be affected with infantile or early-onset FSHD, presenting with a more severe and rapidly progressive form of the disease.

The name facioscapulohumeral muscular dystrophy reflects the stereotypical pattern of muscle groups that are affected: facial muscles (facio), muscles that control the shoulder blades (scapula), and muscles that control the upper arm or humerus. The name is somewhat of a misnomer, however, as abdominal, hip girdle, and lower limb muscles are also frequently involved. A hallmark of FSHD is that muscles are affected asymmetrically and focally, with some muscles such as the bicep or tibialis anterior being more prone to weakening, while adjacent muscles may remain unaffected. Facial muscle weakness may result in an asymmetric or flattened smile, difficulty making facial expressions, and incomplete closure of the eyelids. Affected infants may have sucking difficulties. The shoulder blades may protrude because the muscles which are attached to the scapulae are weakened. Scapular displacement interferes with the ability to raise the arm above

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2 Deenen JC, Arnts H, van der Maarel SM, Padberg GW, Verschuuren JJ, Bakker E, Weinreich SS, Verbeek AL, van Engelen BG. Population-based incidence and prevalence of facioscapulohumeral dystrophy. *Neurology*. 2014 Sep 16;83(12):1056-9.

3 Nguyen K, Robin JD. Facioscapulohumeral Muscular Dystrophy—a Tale of Heterogeneity and the Power of Clinical Assessments. *JAMA Netw Open*. 2020;3(5):e205004.

4 Preston MK, Tawil R, Wang LH. Facioscapulohumeral Muscular Dystrophy. 1999 Mar 8 [updated 2020 Feb 6]. In: Adam MP, Ardinger HH, Pagon RA, Wallace SE, Bean LJH, Stephens K, Amemiya A, editors. *GeneReviews*® [Internet]. Seattle (WA): University of Washington, Seattle; 1993–2020.

5 Wagner KR. Facioscapulohumeral Muscular Dystrophies. *Continuum* (Minneap Minn). 2019 Dec;25(6):1662-1681.



shoulder height. Additionally, weakness of the arm muscles themselves may further limit a person's ability to lift the arms. Other muscles such as those of the pelvic girdle, abdomen, back, and lower limbs may also be affected, leading to frequent falls and difficulty climbing steps, rising from a chair, or standing up after a fall. The weakened muscles typically result in chronic pain, difficulty with mobility and ambulation, impaired function, and psychological distress. Infantile or early-onset FSHD may also be associated with retinal abnormalities, hearing loss, and increased risk of pulmonary insufficiency.

Diagnosis of FSHD requires both clinical and genetic assessments. Genetic testing involves mapping of chromosome 4. Near the tip of this chromosome is a region with multiple copies of a short nucleotide sequence called *D4Z4*. Most people have 11 to 100 copies of *D4Z4*; between 1 and 10 repeats of this gene sequence leads to expression of a gene called *DUX4*, which leads to muscle cell damage. About 95% of FSHD patients have a contraction of the *D4Z4* region on chromosome 4, which in combination with a permissive allele (4qA) allows *DUX4* to be expressed. This aberrant *DUX4* expression is thought to be the underlying pathogenic mechanism that causes FSHD. About 5% of patients have a different genetic anomaly: mutations in other genes such as *SMCHD1* and *DNMT3B*, which result in hypomethylation of the *D4Z4* locus on chromosome 4 and, together with the permissive 4qA allele, cause *DUX4* expression. Thus, both genetic types of FSHD converge on a common pathway: *DUX4* expression with its downstream toxic effects on muscles.

There has recently been progress in developing treatments that inhibit *DUX4* expression using small molecules or by targeting *DUX4* mRNA.<sup>6</sup> Nonetheless, at this time there is no cure for FSHD and no pharmacological treatments to prevent or stop the muscle wasting and weakness that characterize the disease. Management typically involves aquatic, physical, and occupational therapy; exercise; and bracing – particularly ankle-foot-orthoses (AFOs) for foot drop, and abdominal binders for abdominal laxity. Artificial tears during the day and eye ointment at night may help prevent corneal abrasions. Surgery for scapular fixation may reduce pain, improve appearance, and increase arm range of motion, but this procedure is appropriate for only select patients and comes with the risks of any major surgery.

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6 Lim KRQ, Maruyama R, Echigoya Y, Nguyen Q, Zhang A, Khawaja H, Chandra SS, Jones T, Jones P, Chen YW, Yokota T. Inhibition of *DUX4* expression with antisense LNA gapmers as a therapy for facioscapulohumeral muscular dystrophy. *PNAS* Jul 2020, 117 (28) 16509-16515.





## 4. Meeting overview

This meeting was designed to systematically gather patient and caregiver perspectives on FSHD and available treatments. The information from this meeting is assembled in this report, which will be shared with the FDA to inform the agency's review of drug development. It will also be broadly available to the FSHD community and other drug development stakeholders.

Prior to the FSHD EL-PFDD meeting, a survey was conducted with people who have FSHD and their caregivers. The survey helped to determine the meeting content and the selection of panelists who would speak at an in-person meeting (originally planned for April 21, 2020). Because of the COVID-19 pandemic, the in-person meeting was converted to the virtual meeting held on June 29, 2020.

Mark Stone, president and CEO of the FSHD Society and co-moderator for the meeting, welcomed participants to the webcast. Additional opening remarks were provided by Paul Melmeyer, director of regulatory affairs, from the Muscular Dystrophy Association.

The meeting format was then introduced by the meeting moderator, James Valentine, JD MHS, an attorney with Hyman, Phelps & McNamara, P.C. Mr. Valentine previously worked at the FDA, where he helped launch the PFDD program and has helped guide many disease-related organizations in their planning of EL-PFDD meetings.

The FDA's commitment to rare disease drug development and the value it places on understanding the patient experience of diseases such as FSHD, as well as patients' perspectives on benefits and risks of treatments, were discussed by Michelle Campbell, PhD, senior clinical analyst for Stakeholder Engagement and Clinical Outcomes for the Office of Neuroscience, Center for Drug Evaluation and Research (CDER), FDA. Dr. Campbell stated that EL-PFDD meetings such as this provide the FDA with the unparalleled opportunity to hear directly from patients about what matters most to them in the development of medical products.

Kathryn Wagner, MD PhD, director of the Center for Genetic Muscle Disorders at the Kennedy Krieger Institute in Baltimore, MD, and professor of neurology and neuroscience at the Johns Hopkins School of Medicine, provided a medical overview of FSHD.

Following Dr. Wagner's presentation, patients took center stage in two sessions moderated by Mr. Valentine. The first session of the virtual meeting explored patient and caregiver experiences of living with FSHD and its impacts on daily life. The second session focused on how people with FSHD manage their disorder, and their hopes and preferences for future treatments. In each session, a panel of patients and caregivers, selected to represent a range of experiences, presented brief summaries of their experiences. These panels were followed by a moderated audience



discussion in which participants shared their experiences and preferences in real time through comments online or over the phone. A preselected panel of five members of the FSHD community joined through Zoom to provide additional perspectives on the submitted questions.

To start each of the two sessions, all participants in the webcast were asked to respond to a series of questions using a web-based polling technology. This technology enabled immediate capture and display of responses through the webcast. Patients and caregivers were able to participate in remote polling using their telephones and computers. The first set of polling questions collected demographic information about the patients and caregivers participating in the meeting. Only individuals living with FSHD, their direct caregivers, spouses, siblings, parents, or other loved ones were asked to submit responses. For the questions about symptoms, impact of those symptoms, treatments used, etc., caregivers and loved ones were asked to answer on behalf of the patient they care for. While recognizing that FSHD impacts caregivers and family members as well as the person who has the disease, we set our goal for this meeting to better understand the effects of FSHD upon the patients themselves.

The meeting concluded with a meeting summary provided by Larry Bauer, RN MA, Hyman, Phelps, & McNamara, and former member of the FDA Rare Diseases Program, and closing remarks from Mark Stone.

During the 30 days following the meeting, participants and other members of the FSHD community were invited to submit additional written comments by email or on the FSHD Society website. Meeting organizers reached out to several families who had experienced severe manifestations of FSHD but had not participated in the meeting to ensure that their views were adequately represented.

Comments from panelists, other community members, and participants in the webcast, whether voiced during the meeting, submitted in response to the survey and polling questions, or submitted in writing, are compiled in this summary “Voice of the Patient” report.

## Pre-meeting survey

Nearly 600 people responded to the pre-meeting survey. Of those who responded, 92% were individuals with FSHD; about 10% were parents or legal guardians of a child with FSHD, and 4% represented adults with FSHD. (Because FSHD is inherited, individuals can have FSHD and also be a caregiver or guardian for affected family members.) They lived all around the world, with more than 60% of survey respondents residing in the United States and about 26% in Europe or the United Kingdom. Survey respondents also hailed from Canada, Asia, Australia, New Zealand, Central and South America, and Africa.

**In terms of age distribution:**

- 59% were 50 years of age or older
- 17% between 40 and 49
- 10% between 30 and 39
- 11% between 18 and 29
- 3% were younger than 18

Females outnumbered males (54% versus 46%). Ninety-three percent of respondents identified as White, 4.5% as Asian, and 2% as Latin American. Only one respondent identified as Black and one as Native American or Alaskan. Approximately 60% had been diagnosed more than 10 years ago.

Nearly 60% of survey respondents had received a genetic diagnosis, about 27% were not sure, and 16% said they were never tested.

**Of those who had a genetic diagnosis:**

- 86% had Type 1 FSHD
- 9% had Type 2
- 3% had both Type 1 and Type 2
- 2% had inconclusive results

The survey asked a series of open-ended questions in order to capture the full range of patient experiences with FSHD. These questions asked about the impact of symptoms, what worries patients the most, current treatments, and what would be most meaningful and important for future treatments. Survey responses were subsequently categorized to create in-meeting polling questions that could be answered in real time on the day of the meeting.

**Real-time polling**

During the webcast, 307 participants logged on to the polling app and responded to some or all of the polling questions. About half of the webcast participants provided demographic information and general information about their disease. Seventy-four percent were individuals living with FSHD; 26% were parents or caregivers. Eighty-five percent were from the US (across all regions). Other areas represented included Europe, the UK, Canada, Central or South America, Asia, and Australia or New Zealand. Fifty-two percent were female.



### **Age distribution of polling respondents**

- 64% were over age 50
- 9% age 40 to 49
- 9% age 30 to 39
- 10% age 18 to 29
- 7% younger than 18

### **Disease characteristics of polling respondents**

Among those who answered polling questions about their disease, 63% were diagnosed more than 10 years ago, 11% between five and 10 years ago, and 25% within the past five years.

Regarding their diagnosis, 81% said they had a genetic diagnosis, 10% were never tested, and 8% were not sure if they had received a genetic diagnosis.

#### **Of those who had been genetically tested:**

- 94% received a diagnosis of Type 1 FSHD
- Fewer than 3% received a diagnosis of Type 2
- 1% received a diagnosis of both Type 1 and Type 2
- 1% had inconclusive results



## 5. TOPIC 1:

### **Symptoms of FSHD and daily impacts that matter most to patients**

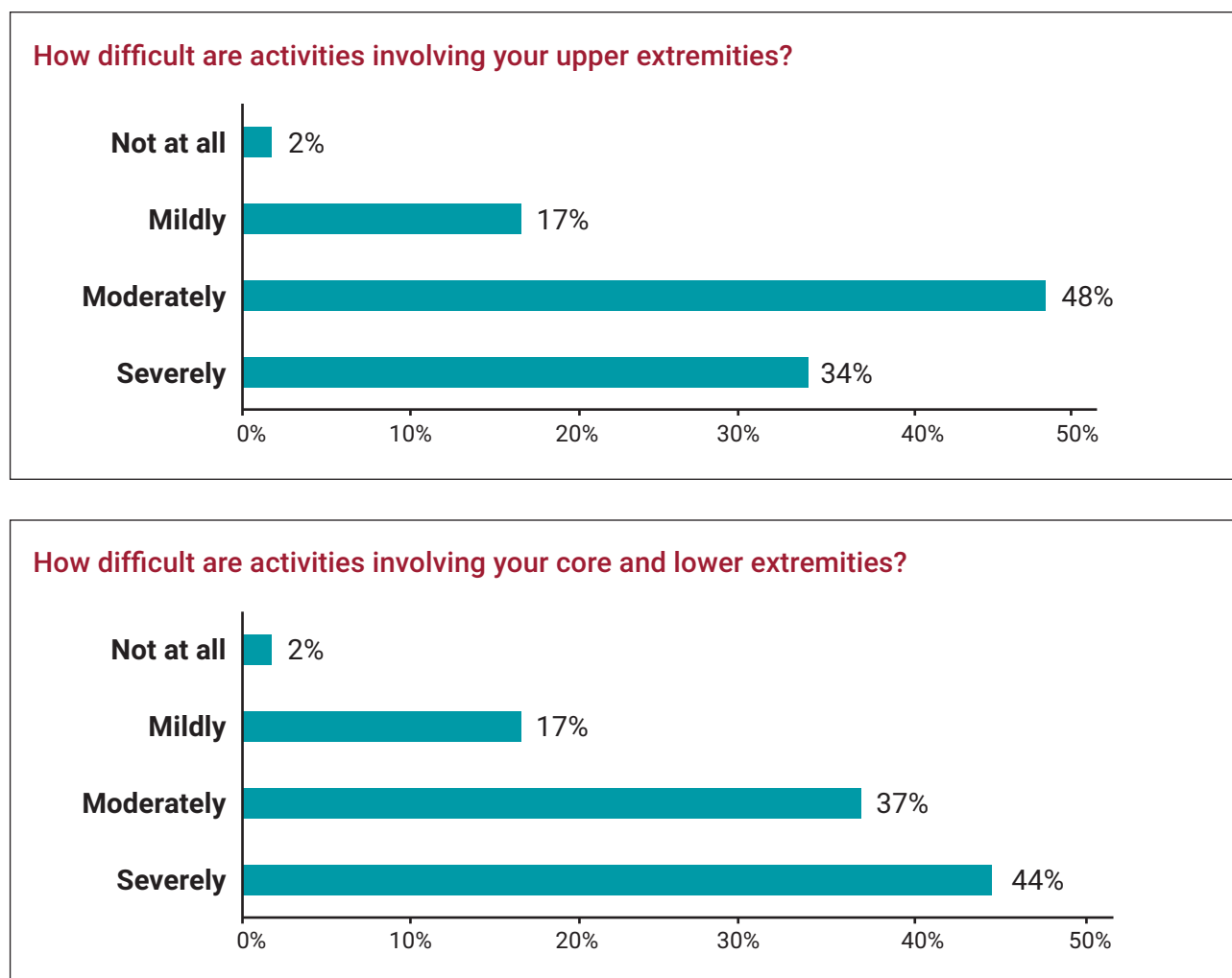
The polling questions asked respondents about the difficulties they experienced involving upper and lower extremities, and to select from a list the symptoms that have most impacted their daily lives. As described earlier, symptoms vary widely from person to person and typically progress over time.

#### **Muscle weakness and other symptoms**

As shown in Figure 1, these data indicate that most people with FSHD report moderate to severe difficulties involving both upper extremities and core/lower extremities, with somewhat greater severity in the lower body. The narratives shared by workshop participants provided additional insight into how muscle weakness can make even the simplest tasks difficult.



FIGURE 1. LEVEL OF DIFFICULTY DOING ACTIVITIES INVOLVING UPPER AND LOWER EXTREMITIES



### SYMPTOM: UPPER AND LOWER EXTREMITY WEAKNESS

One 27-year-old woman said, *“I began holding my wrist with my other hand when eating.... I’ve given up writing with paper and pen because I have low dexterity.”*

A 67-year-old recently diagnosed woman said, *“I almost can’t get a fork to my mouth because of my arm limitations.”*

A 50-year-old man added, *“Forget doing laundry; the clothes are too heavy. Forget making the bed; I’ll end up on the ground. Forget loading the dishwasher; the plates are too heavy, and forget carrying a glass of water; I don’t have the balance.”*





Patients also described how progressive upper body weakness has impacted other aspects of their lives: *“I wasn’t able to pick up my kids out of their crib or throw them in the air like many of my friends did with their kids. I had to sit down as my wife handed them to me, just like she did with their great-grandmother who was fragile and weak,”* said one man.

A 38-year-old woman added, *“I have lost so much functionality that I cannot care for my kids without support. I cannot carry my kids, lift them out of the crib, and my right arm has atrophied so quickly that I can only type or use my phone for limited periods of time.... Doing my hair can take up most of my energy for the day or even my makeup, so I do not go out very often because of the effort, and my self-esteem has suffered.”*

### **SYMPTOM: FACIAL WEAKNESS**

Weak facial muscles may also have a profound impact on a person’s overall health and quality of life by affecting their speech, ability to eat, and smile.

The mother of an 11-year-old girl diagnosed with early-onset FSHD at age eight wrote, *“She had hearing loss and a complete lack of expression in her face. She couldn’t raise her eyebrows or smile, and her speech was unintelligible.”*

The mother of a 17-year-old woman with early-onset FSHD diagnosed at age three wrote, *“She has sleep apnea due to weak face and neck muscles, so when she lies down her upper airway is compromised. She uses a nighttime BiPAP machine.”*

The mother of a seven-year-old girl with early-onset FSHD wrote, *“Eating has always been a major challenge, we think because her weaker muscles in her face and mouth just make it harder, so we end up feeding her about half of every meal, to make sure she gets the calories she needs.”*

A 27-year-old woman said, *“Chewing with my mouth closed became impossible due to the facial weakness.... My smile and facial expressions weaken with every year.”*

Another woman who early in her career gave frequent presentations described how weak facial muscles have made this much more difficult: *“Each word I use must be weighed. Will it be difficult to pronounce? Is there another word that would convey the same emotion in a meeting that might be easier for me to use? When I’m giving a presentation, I can see the faces in the audience, as they try to understand me.”*



## SYMPTOM: CORE WEAKNESS AND RESPIRATORY PROBLEMS

Weakened core muscles contribute to many problems including difficulty breathing. Although only a minority of workshop participants reported respiratory problems as having the most significant impact on daily life, for people with severe or late-stage FSHD, the consequences of respiratory problems can be serious, including sleep disordered breathing, respiratory insufficiency, and acute respiratory failure.<sup>7</sup>

The daughter of a woman with FSHD who died at age 76 wrote, *“Because of her weak core due to FSHD, my mom wore a corset to keep her back straight and stable for as long as I can remember.... The corset was tight so she could stay straight, but it also impacted her lungs and made it hard for her to cough and clear her lungs. She was at high risk for choking and pneumonia, especially during allergy and cold/flu season. More often than not, she ended up in the hospital if she got a cold so they could help clear her lungs for her and prevent pneumonia.”*

The mother of a 17-year-old woman with infantile FSHD wrote, *“She has severely weak core muscles and therefore has difficulty taking a deep breath, and is unable to effectively cough.”*

The mother of a seven-year-old girl with early-onset FSHD wrote, *“She has been hospitalized multiple times for pneumonia and respiratory illnesses, so much that she now has permanent damage to her lungs (bronchiectasis, or lung disease).”*

One man wrote, *“My body is effectively completely paralyzed due to FSHD, and I am fully ventilator dependent, but one of the most debilitating aspects to me of my own muscle deterioration is having lost my speaking voice.... My mind remains active and vibrant – I have a doctoral degree in mathematics – yet I can no longer express myself verbally in most situations. I cannot mime or point to compensate for my absent voice, nor even write my thoughts on a pad of paper, because FSHD has stripped me of most voluntary muscle movement. My communication difficulties have even put my life in danger. I have been in the hospital for reasons unrelated to FSHD when my ventilator has accidentally become disconnected, yet I was unable to communicate my difficulty breathing.”*

## SYMPTOM: COGNITIVE AND SENSORY IMPAIRMENTS

While central nervous system involvement is not a core part of the disease, cognitive impairment has been associated with FSHD.<sup>8</sup>

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7 Trucco F, Pedemonte M, Fiorillo C, Tacchetti P, Brisca G, Bruno C, Minetti C. Respiratory pattern in a FSHD pediatric population. *Respir Med*. 2016 Oct;119:78-80.

8 Mah JK, Chen YW. A Pediatric Review of Facioscapulohumeral Muscular Dystrophy. *J Pediatr Neurol*. 2018;16(4):222-231.



The mother of a seven-year-old girl with early-onset FSHD wrote, *“She has significant cognitive, speech, fine motor, and gross motor delays.”*

The mother of a seven-year-old boy with early-onset FSHD wrote that her son is *“non-verbal and cognitively much younger in age.... He was born with bilateral sensorineural hearing loss.”* He is also totally blind in both eyes, the result of a condition in which the blood vessels behind his retinas leaked fluid, causing retinal detachment.

## PROGRESSION OF SYMPTOMS

Like the symptoms themselves, the rate of progression is highly variable over the lifespan. Progression may start early in life and proceed unabated, while other people may experience many years of minimal progression followed by an abrupt change to faster progression.

A 27-year-old woman said, *“Twenty-two was the last time I used a regular stall bathroom because I couldn’t stand without grab bars. [Age] 23 was the last time I drove because I couldn’t pick my foot up off the pedals. At 24 I purchased a power wheelchair because the scooter wasn’t sturdy enough for me. Twenty-five was the last time I stood to take a shower because I fell often. Twenty-six was the last time I walked and also the last time I stood on my own.”*

A 54-year-old man who was diagnosed at age 17 said he thought for a long time that he had a milder and slower-progressing form of FSHD. *“At 50 years old, it was like a light switch that was flipped – the increased pain in my legs. Now it’s transitioned into my sleep and it’s affecting my sleep. I am losing my ability to walk. I’m a musician but I’m losing my ability to play music.”*

A 60-year-old man diagnosed with FSHD1 at age 32 said, *“The rate of progression of this disease for me, over the last three years, has been just downright frightening.”*

The mother of an 11-year-old girl diagnosed with early-onset FSHD at age eight said her daughter *“... can barely run anymore. She can’t lift both arms overhead. She struggles to walk up the stairs.... One day she was fine and the next her shoulder was slouched and had lost muscle ... overnight!”*

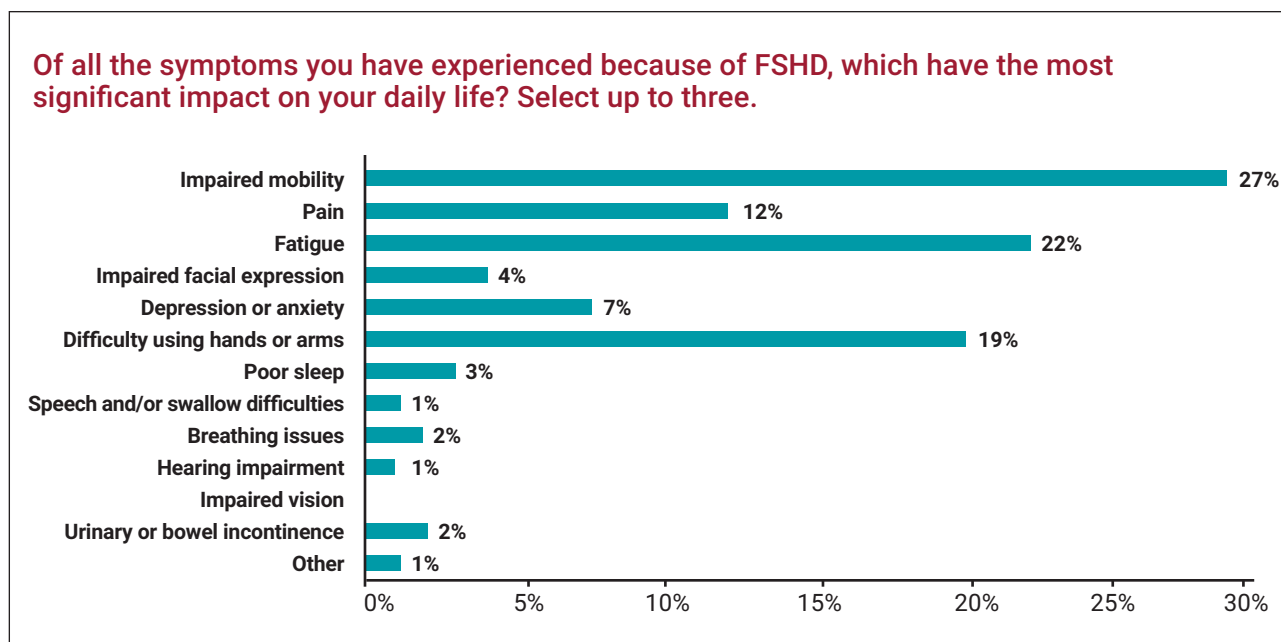
## Consequences of FSHD symptoms

It is not just the weakness per se that contributes to the limitations in performing day-to-day tasks, but its consequences, including impaired mobility, fatigue, and difficulty using hands and arms. As shown in Figure 2, these three symptoms were reported to have the most significant impact on daily life. Mobility impairments and fatigue also contribute to a high rate of falls among people with FSHD. A subset of FSHD patients experience pulmonary insufficiency due to weakening of



muscles involved in exhalation, resulting in a weakened cough and hypercarbia (elevated blood carbon dioxide). These conditions increase the risk of pneumonia, respiratory failure, and premature mortality in this subset.

FIGURE 2. SYMPTOMS WITH THE MOST SIGNIFICANT IMPACT ON DAILY LIFE



## CONSEQUENCE: IMPAIRED MOBILITY

A 50-year-old woman said, *"In my freshman year in high school I was a cheerleader. By my sophomore year, I couldn't stand up from the ground by myself."*

Another woman said when she was diagnosed at age 13, she thought she would be okay as long as she could continue to ski. She skied for the last time at age 17, and now she struggles to climb stairs. *"I walk with significant gait and trip a lot because of foot drop, and I can no longer get up from the floor when I fall. And recently on a windy day, I fell over and had to crawl for 30 meters to find a step where I could hoist myself up. It was degrading and humiliating. It's devastating to have lost so much at age 26."*

A 27-year-old woman described how at age 14 she sensed something had changed during her typical three-mile run: *"I couldn't propel my legs forward like they used to, and a mile in I collapsed on the cul-de-sac crying, my favorite thing stripped out from under me. I gave it another year-and-a-half, and 10th grade flag football was the last time I ran."* Her difficulties living the normal life of a teenager continued throughout high school and into college: *"First, it was grabbing for a handrail, then it turned into using my right leg only to ascend stairs one step at a time. Falling downstairs became a weekly thing. My friends were talking about kissing boys, and I was worried about not*



*tripping and falling on the graduation stage steps. My gait and lordosis [curvature of the spine] became more pronounced, and I ignored the mean comments everywhere that I went. Sophomore year of college, I fractured my skull after tripping due to the foot drop. Eleven days in the hospital and a tender spot on the back of my head for the rest of my life."*

## CONSEQUENCE: FATIGUE

Mobility issues and muscle weakness frequently result in severe fatigue, which may impact many aspects of a person's life.

A 50-year-old woman said, *"I work on computers. Just putting my hands out to type and to do keystrokes can get exhausting, and it's like, 'Oh, great, what's the shortest way I can convey what I need to say without having to type an extensive amount of time?"*

Weakness and fatigue impact other aspects of patients' lives as well. Said one woman, *"I miss the role of being an active member of my community, whether it's in a volunteer position for a school, or serving on committees, or serving on boards and the like, and one of my issues is that I can no longer stand at a podium."*

## CONSEQUENCE: FREQUENT FALLS

Weakness and fatigue also lead to increased falls among people with FSHD.

A man said, *"Playing golf, a game I loved, was no longer an option because my quads were not strong enough to support my weight. Then came the falls, which are often unannounced and terrifying. A strong breeze can knock me over, uneven pavement will cause me to trip and fall, being bumped into will knock me off balance and I'll go down, or my knees will just unexpectedly buckle which will put me on my back... Other falls have resulted in a pulled groin, bloody face, torn meniscus, bruised hip, bruised elbow, broken rib, broken finger, scuffed up knees, black eye, and intense headaches from hitting my head on the ground. Now at age 50 walking is limited to a few steps inside my house, and at this point it's easier to list the muscles I still have versus the ones I've lost."*

One woman said, *"I fall a lot, I always have bruises on me, I always have cuts and scrapes on me and I usually have something sprained. I think that's one of the hardest things.... I have times where, when I'm going through periods of more muscle loss, or a lot of stress, or a lot of fatigue, or there's just a lot going on, I'll notice that I fall more during those times, but I usually fall at least a couple of times a week.... I've fallen several times where I'm alone and I'm in the rain, and I had to sit outside in the rain by myself, with bloody knees because I can't get up by myself. So I just sit there for an hour until someone comes along."*



A man participating in the Zoom panel said, *"I fell leaving the office because I was fatigued after a full workday. I stumbled in the office hallway, fell awkwardly and snapped my femur in two. And I dedicated a whole year trying to get back to walking. That was not successful, and I started using a wheelchair."*

## CONSEQUENCE: PAIN

The combination of weakness and falls may also result in an extraordinary amount of pain in people with FSHD, and many patients reported pain as their most severe and impactful symptom.

Pain may result from physical problems that develop over the course of the disease. For example, one woman said, *"There's the spine pain, because of the curvature of the spine; I get the lower back pain constantly."*

One woman described how weakness and pain changed over the course of her disease as different muscle groups became affected: *"When I was walking [I was] exhausted from walking so much and then being in a car and my back hurting, and things like that. It's difficult in a different way now because now my arms are so severely impacted.... So the pain from walking in my back and my legs doesn't exist anymore, but now my shoulders and my arms hurt, or just now it's the breathing issues that I have make it difficult because now I'm exhausted just from sitting up all day."*

The pain experienced by children with early-onset disease can be particularly excruciating. In a written comment submitted online, a mother said, *"My son no longer has the muscle strength to hold up his head easily. He has daily neck and back pain due to this difficulty.... He takes lots of painkillers, yet the pain is severe and daily. Sometimes the pain is so bad that he vomits."*

Pain also becomes substantially worse over time for many people with FSHD. A 58-year-old man said, *"I'm in constant pain from all the wear and tear put on my body from having to compensate for the lack of muscle, not to mention all the falls. Instead of living my golden years, it's the broken years."*

Another man said, *"Nowadays pain is a constant companion; my lower back hurts from sitting so much, and the extreme hyperextension in my left leg results in knee pain after walking only a handful of steps. Being able to stand stationary relieves my back pain, but eventually my right leg goes numb and I need to sit down. Finding comfort is an ongoing battle."*

A 57-year-old woman diagnosed at age 18 added that in the last three or four years, *"The pain has really become so bad for me, my ankles, my lower back, my neck.... As soon as I get up in the morning, I start walking for a half an hour. I have to sit down, my back hurts, my ankles hurt, and that just puts a damper on the rest of the day."*





## CONSEQUENCE: DEPRESSION AND OTHER PSYCHOLOGICAL DISORDERS

The daughter of a woman with FSHD who died in 2016 wrote, *"The constant planning and re-planning required to make sure your body does not fail you or you have the access you need are exhausting, and the anxiety that goes with it sucks the life out of you. You find it easier and less stressful to avoid it all and just stay home. The more you stay home, the more isolated you become, and depression sets in."*

The father of two adult daughters with FSHD said of his younger, less affected child, *"She's very, very scared because she has seen the decline in her sister's condition over the years. So scared, in fact, that we fear she tends to put distance between herself and the rest of us. Anxiety and depression are also her constant challenges."*

## CONSEQUENCE: PULMONARY DISEASE AND PREMATURE DEATH

A 59-year-old man wrote, *"Respiratory issues such as sleep disordered breathing, insufficiency, and acute respiratory failure are of issue in FSHD patients. I've experienced both long-term chronic progressive issues as well as acute respiratory failure due to CO2 retention. I now use a ventilator."*

A woman recalled her late mother's ordeal after weakness in breathing muscles led to a hospitalization: *"My mom had a bad cold and was having difficulty coughing to clear her lungs. She went to the hospital, and their pulmonologist told us that due to her blood pressure dropping so low when she coughed and her weakness from FSHD, she would die without a tracheostomy, which they performed against her wishes. She was put on a ventilator and then sent to a horrible rehab center for six weeks to try to wean her from the ventilator. The rehab center gave her no support, and because my mom could not move without assistance, she was completely neglected while she was there. The whole family fought to get her released, but it was not possible until her six weeks of Medicare payments ran out – and only then was she allowed to return home with the ventilator and tracheostomy she never wanted.... Finally, after several years of hard work, my mom was able to convince a pulmonologist to remove the tracheostomy. My mom told me later that the tracheostomy was never needed, and if we only had someone on our side that could explain FSHD, this would have never happened."*

A mother who has two children with FSHD, one who had early onset, recalled that her son *"... was diagnosed with infantile FSHD, relatively rare and much more severe; he was in a wheelchair permanently by age 12 with profound muscle weakness throughout his body.... He completed college in early 2009 with a bachelor's degree in computer science.... In 2012 [he] started to weaken even more, and due to lung issues almost died later that year. He was on oxygen, and*



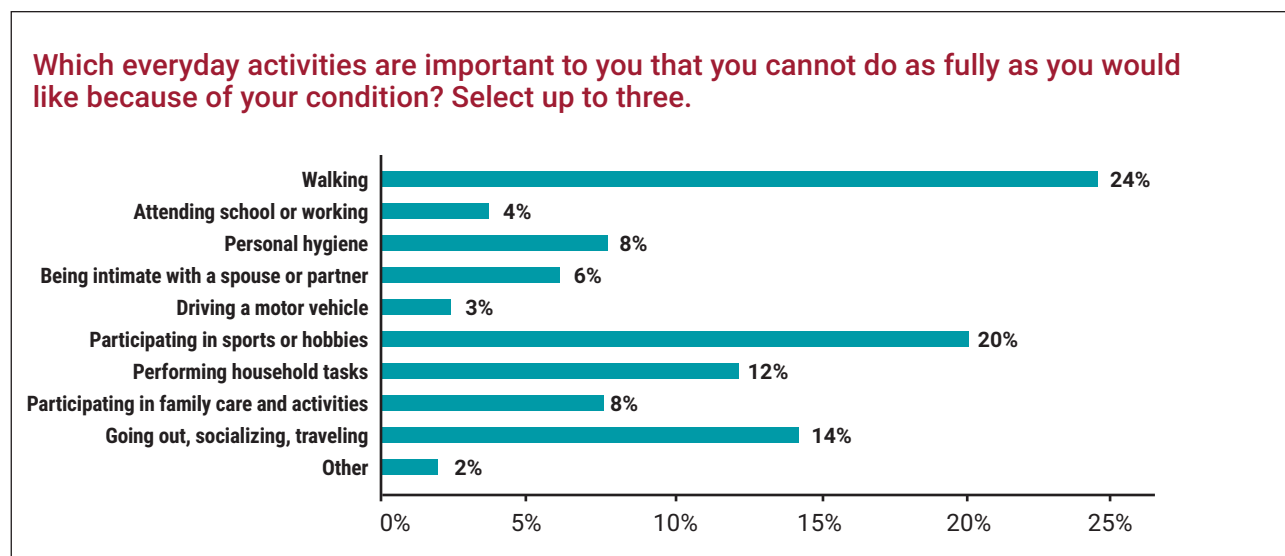
*we placed him on hospice, weighing in at 66 pounds. Even though [our son] made a remarkable recovery, 'graduating' from hospice, he required full-time support from us.... In early 2017, he contracted a common cold and died a week later at the age of 29 due to effects of pneumonia."*

A woman who, along with her brother, inherited FSHD from their mother said, *"No matter how hard my mom fought, FSHD stole her independence, turned her into a quadriplegic, and when she got that last lung infection, FSHD had destroyed so much of her body that there was nothing left for her to fight the infection with. My mom fought this disease with everything she had every day of her life, but in the end, FSHD won and her body failed her."*

## Impact on daily life

Participants were also asked which daily activities that are important to them are most impacted by FSHD. As illustrated in Figure 3 and described in the quotes below, walking and participating in sports or hobbies were selected most frequently, reflecting the impact of impaired mobility, weakness, and fatigue. Other highly impacted activities include going out, socializing, traveling, and performing household tasks.

FIGURE 3. DAILY ACTIVITIES MOST IMPACTED BY FSHD



## IMPACT: WALKING/MOBILITY

A 58-year-old man with FSHD said, *"While you are walking, you're always looking on the ground for anything that might throw off your balance: sticks, pebbles, cracks, anything uneven. When you walk into a room, you have to survey all your surroundings and strategically plan where you can sit, usually by a desk so you can push off with one muscle, pull with another to stand up, or if you fall, how you will get back up."*



A man with a mild form of FSHD described the effects on his two more severely affected daughters: The older daughter (now ~35) has *"... severe difficulty walking, and even for short distances she needs a wheelchair.... She can barely go up steps, and she has long been unable to visit the second floor of our house. She has great difficulty sitting or getting up from a chair without falling.... If she falls, she must crawl to a bed or other furniture to pull herself up."*

## IMPACT: PARTICIPATING IN SPORTS OR HOBBIES

The mother of a 14-year-old boy with FSHD described how when he was 10, she noticed he could not do a sit-up. *"Although he had played sports since he was five years old, his endurance was decreasing, and he wasn't able to keep up with his friends. By age 11 he chose not to sign up for basketball or baseball. Since then he joined Boy Scouts and middle school band but was unable to pass the swim test at Boy Scout camp, which deprived him of some swimming activities. He cannot walk with a heavy backpack for an overnight camping trip, and it is a significant challenge for him to go on a five-mile hike with his fellow scouts. As he is unable to purse his lips, he was limited on the instruments he could select to play in band. However, he determined that he could play percussion, and he has taken a liking to this new hobby."*

A 50-year-old man said, *"I liked to play sports, work up a sweat, go for a hike. Now most of the things I'm passionate about have been taken from me; now I get around in a scooter, which has been a major adjustment both physically and mentally."*

A woman on the Zoom panel said, *"I used to be a golfer, and I had to give that up. I'm an artist, and I can no longer hold the brush to the canvas. People say, 'Well, be inventive.' I come from a multiple-generation family with FSHD, of cousins and uncles, and my father and grandparents. I have come to expect and learn how to be inventive and create adaptive equipment. There comes a point when none of that helps."*

## IMPACT: WORKING

A 50-year-old woman said, *"The neurologist that diagnosed me was pretty blunt. He said, 'Change your major, get a desk job. Don't overuse the muscles and think twice about having kids....' I was a costume designer and ... that meant that I was on my feet, 10 to 13 hours a day, six days a week, and the day off that I had, I spent recuperating where others were off partying, doing fun things. I was just pretty much watching TV, trying to recuperate, getting ready for the next week."*



## IMPACT: GOING OUT, SOCIALIZING, TRAVELING

A 27-year-old woman described grieving for different things at different stages of life. *"I'm currently in this stage where going out is important, being a young adult, and you have to weigh what's important and balance things. So if you're too exhausted from trying to take care of yourself throughout the entire day, when your friends want to go out at a bar at night or go hang out, that may not be something that you can do, because you're too exhausted ... even traveling or being on a plane or in a car for an extended period of time, maybe it's just being too exhausted from sitting up the entire time..."*

A 50-year-old woman said, *"I have not walked or stood since I was in my mid-30s. Can't do it. It's impossible. So to go out and be in the community and society, I have to plan everything. I don't drive anymore, so, okay. Is there a bus or transportation? Does the restaurant have enough room to accommodate [a wheelchair], is the bathroom accessible? I have to define and plan ahead for every situation, even going to the grocery store."*

## IMPACT: SELF-CARE

A woman said, *"The activities of daily living, with pain and an inability to raise your arms, is indescribable. Imagine, if you will, you were tied up by a burglar and they took a strap and they wrapped it around your entire upper body and both arms and your trunk, and you could not lift your arms more than six to maybe 12 inches, and then you want to scratch the top of your head, or you want to brush your teeth, or you want to reach a shelf. And the effort that you have to exert ... in order to even get a modicum of movement towards your upper body, towards grooming, brushing teeth, dressing, putting on a shirt, showering, is so exhausting, that that's where the fatigue and the mental challenge comes in."*

Another woman added, *"Just to be able to put on a shirt in the morning, I have to place my elbows on a desk or countertop to put on a shirt. To wash, I have to put an elbow on the wall to get to wash the hair, to get the hands up."*

## IMPACT: LOSS OF INDEPENDENCE

The mother of a 17-year-old girl with early-onset FSHD wrote, *"Pain and the need for a companion caregiver keep her from doing a lot of extracurricular activities. Simple things such as going to dinner with her fellow graduates isn't something she can do without someone who can assist her or who knows how to manage someone who is choking."*



The mother of a seven-year-old multi-disabled boy with infantile FSHD wrote, *“Over time, I have come to accept that he will never be independent, have friendships, or regain vision. Maybe he will never talk.”*

## IMPACT: EMOTIONAL

A 50-year-old man said, *“The physical and mental pain is accelerating, and my optimistic nature is being tested like never before.”*

A 50-year-old woman added, *“It’s like a cyclical grieving process.... You stabilize, and then you lose something else, and it’s just like losing another loved one.”*

The father of two daughters with FSHD said of his older daughter, *“Despite her enormous emotional strength, she sometimes has panic attacks and struggles with loneliness and depression.”*

## IMPACT ON FAMILY LIFE

The mother of a 17-year-old girl with early-onset FSHD said, *“Our small, four-member family has been impacted by this disease in negative as well as positive ways. Nothing we do looks like what a typical family does. We don’t just jump in the car and go somewhere. It takes planning, effort, supplies, a wheelchair-accessible vehicle, and a service dog. We don’t leave her alone for more than 60 minutes in case she needs something. But because of this, we are a very strong family unit; we have a large support network of extended family, friends, and caregiver support; we are patient and thankful for the small, simple things in life; and we take nothing for granted. Each day is a blessing.”*

A 58-year-old man said, *“I made a decision not to have children after watching all of the troubles and heartache my mom and grandfather went through in their battle with FSHD.... Nowadays, I feel not having children was a bad decision; I could definitely use the help from them now. Most of all, I missed out on so very much. Everyone should be able to have the experience of having children and grandchildren, even though I am unable to lift and hug a child.”* He added, *“I lived by myself for 10 years before I was introduced to the lady I live with now. I am so worried that I will lose her because I am unable to make love to her because of the disability.”*

A 54-year-old man described similar losses: *“Six years ago today, I was blessed with my very first grandchild. Recently, we were blessed with two baby boys that I can’t hold now. My first grandchild I was able to pick up and hold. My two boys, I have to wait until they are old enough to walk before I can take them anywhere or be really involved in their lives like I’d like to be.”*



A 61-year-old man diagnosed at age 34 described how he was excluded from a family gathering: *"I guess the problem is that accommodating for somebody with a physical disability, i.e., a power wheelchair, using a power wheelchair, just might be a bit too difficult for some people ... if I were able to go and visit, I would not be able to use the bathroom.... Why can't I go visit my friends? Why can't I go visit my family? Why can't I participate in these activities? And the reason is, in many cases, because you just physically cannot do that."*

A woman from Canada commented that beyond family members being unwilling or unable to make the accommodations needed, the disease impacts those family members as well: *"Spouses have to watch someone that they love decline and deteriorate, and it's not just weeks and months they have to watch this ... the loss is profound. And many times family and friends cannot cope with this emotionally. It is [as] devastating for them to watch as it is for parents to watch their own children decline in this way."*

A 67-year-old woman described a scary situation while watching her granddaughter: *"About two weeks ago, I went to visit her. We were taking a walk and she took off like a rocket at two-and-a-half years old towards the highway.... And I couldn't catch her. I can't run anymore. Luckily, she stopped at the road. It's just beyond devastating."*

Another woman wrote, *"I was always very active, and I have a very active family. It is very hard to be the person who limits our activities. We have to pick shorter hikes, shorter camping trips, and take more breaks on family vacations, etc., because mom can't keep up. It also takes me longer to recover after an active day, which just puts more limits on what we do. What's hard is knowing that this will only get worse. Eventually, our family won't just be shortening a family hike, but maybe not be going at all, or maybe leaving mom at home."*

The mother of a child with infantile FSHD said, *"The simplest outing for [our daughter] has to be thought through versus just being able to go. Nonstop problem-solving, worrying, and strategizing on all our parts just to leave the house.... It takes a lot of time, research, and chatting with many doctors and other patients to come up with a game plan for your own situation. This has been a full-time job on top of the ones my husband and I already hold ... we strive to make sure she has as much of a normal childhood as possible; we have revamped and changed our lives drastically to do what we can to slow progression.... We have revamped our entire way of eating with whole foods and farm-to-table. It's time consuming and expensive, but if it helps just keep her where she is today or slows progression, we're willing to endure."*

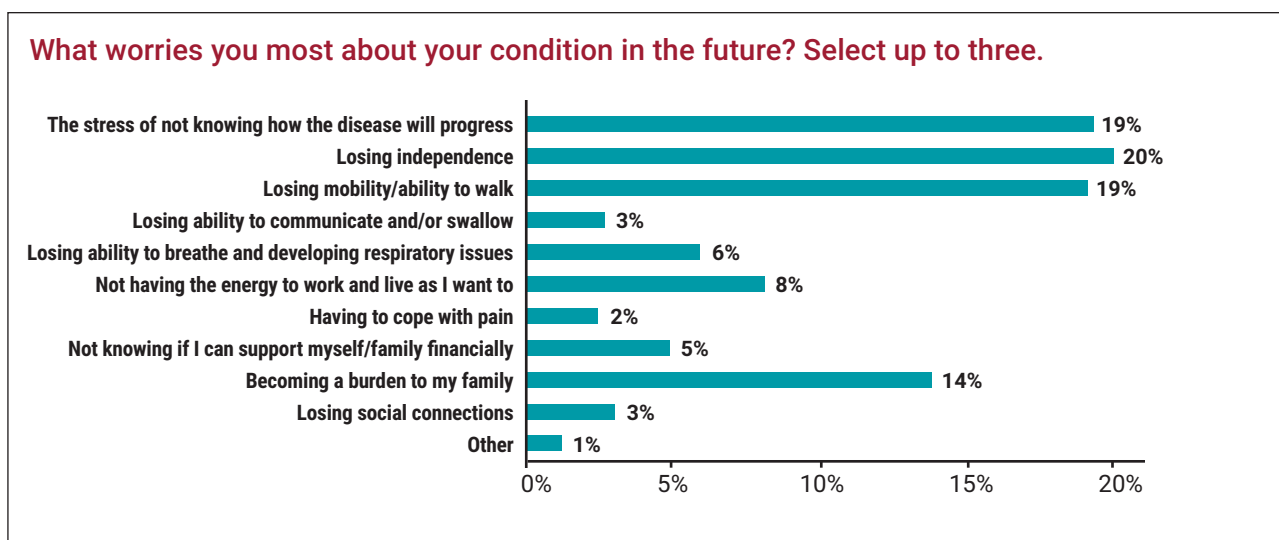




## Concerns about the future

For many people with FSHD, the progression of the disease is relentless and frightening. Some people described a fairly mild disease in early life that shifted gradually to a more severe form later on. Nearly everyone with FSHD worries about the future. Polling responses (Figure 4) indicated that the most prevalent of these worries included losing independence (20%), losing mobility/ability to walk (19%), the stress of not knowing how the disease will progress (19%), and becoming a burden to their family (14%).

FIGURE 4. WORRIES FOR THE FUTURE



## FUTURE WORRIES: LOSING INDEPENDENCE

A 50-year-old man said, *“This disease is wicked and cruel in many ways, but losing my independence is probably the most frightening and helpless feeling I have ever had.”*



A mother said her son is already limited in many activities that his friends enjoy. *“However, it is not too long before more meaningful activities are limited. Normal things like climbing stairs, carrying luggage, and reaching for something out of the cabinet, we take for granted. These limitations are around the corner for him.”*

The father of a 36-year-old woman with FSHD said, *“She’s now living independently and working, with her own car and condo.... We especially worry about how long she will be able to continue to live independently in her condo, or drive her car, or when and how she will have to modify them both substantially.”*

### **FUTURE WORRIES: LOSING MOBILITY/ABILITY TO WALK**

A 27-year-old woman said, *“I’ve fallen by myself many times, and I think one of the scariest things about falling, especially by yourself, is I’m scared that I’m going to hit my head, and I’m scared that someone’s going to find me unconscious. And I’m scared that, one day I’ll fall and I’ll break something, and lose even more mobility.”*

### **FUTURE WORRIES: STRESS OF NOT KNOWING HOW DISEASE WILL PROGRESS**

A 27-year-old woman said, *“I think about the last time I’ll be able to choose solid food because choking occurs. I worry about deteriorating into absolutely nothing but a bag of bones when all the muscle and fat dwindle away.”*

A 35-year-old woman said, *“Waiting for a treatment is scary. I am fearful of what else I will lose while I wait. Will FSHD take mobility from my hands? Will breathing become more difficult? What else will it take away from my family members who are sick?”*

A 44-year-old man said, *“I was diagnosed four years ago, at 40, and it felt like someone started a stopwatch as soon as I heard I had so little time to physically enjoy the things I was enjoying.... The scary thing is, you don’t know how much time you have left. And I understand that that is life in general, but for FSHD it seems like it’s awful quick, and the time you thought you might have, you don’t. It’s abbreviated, because it’s so unpredictable.”*

A 54-year-old man added, *“The constant not knowing as life progresses [how] this disease will progress. It’s just torture.”*



A woman who has lived with FSHD for nearly 30 years and survived breast cancer said she had *“... an amazing oncologist who calmly laid out a plan to beat cancer.... I later realized that having this plan was what got me through this nightmare. He was confident we could beat the cancer. And with his plan we did.... FSHD in some ways is worse than cancer because there’s no treatment, there’s no plan. There’s no guidance.”*

A 60-year-old man said, *“Three years ago, I was jogging. Today I’m having trouble walking. The uncertainty about what’s next is definitely affecting my quality of life.”*

The father of two daughters with FSHD said, *“Any conversation about a permanent wheelchair or a scooter is extremely painful, both because of the fear it creates among all of us and because nobody knows what the progression of the disease might or might not be.”*

For parents of children with early-onset FSHD, worries about progression are constant. Said one mother of her son, *“At the age of 14 he is learning how to accommodate for his muscle weakness. I worry about the changes and modifications he will have to make throughout his high school years, college years, and adult life.”*

For parents who themselves have FSHD, observing the disease in their children can be terrifying. Said one mother, *“Of my three children, two now have a diagnosis, and I watched my third one like a hawk.... My fears for their future feel as though they’ll crush me; both my girls have visible symptoms 20 years before I did.”*

## **FUTURE WORRIES: BECOMING A BURDEN TO FAMILY**

A 58-year-old man said, *“It weighs on me all the time that I am becoming too much of a burden the more the FSHD progresses.”*

A 44-year-old man added, *“How am I going to have income for my family, as my job I used to do I can no longer do?”*

A Zoom participant described the toll FSHD took on his family: *“It was the toll that it took on my mom’s health as a caregiver to my dad that I most noticed growing up. And that haunts me to this day, and one of my biggest worries going forward is the effects of what this disease can have on the people who truly care for us. We want to be independent and be able to handle this disease ourselves. But the fact of the matter is all of our friends and family are very much intimately involved. And my constant fear is the effects that has on them.”*



## 6. TOPIC 2: Managing FSHD

Building on the discussion of the symptoms and their impacts on daily life of people with FSHD, patients and caregivers participating in the PFDD meeting were next asked to discuss in more detail how they manage the disease, including both pharmacological and non-pharmacological treatments, the benefits and adverse effects associated with those management strategies, and their preferences for future treatments. Among the chief drawbacks of current therapies identified by meeting participants are limited effectiveness, side effects, and costs.

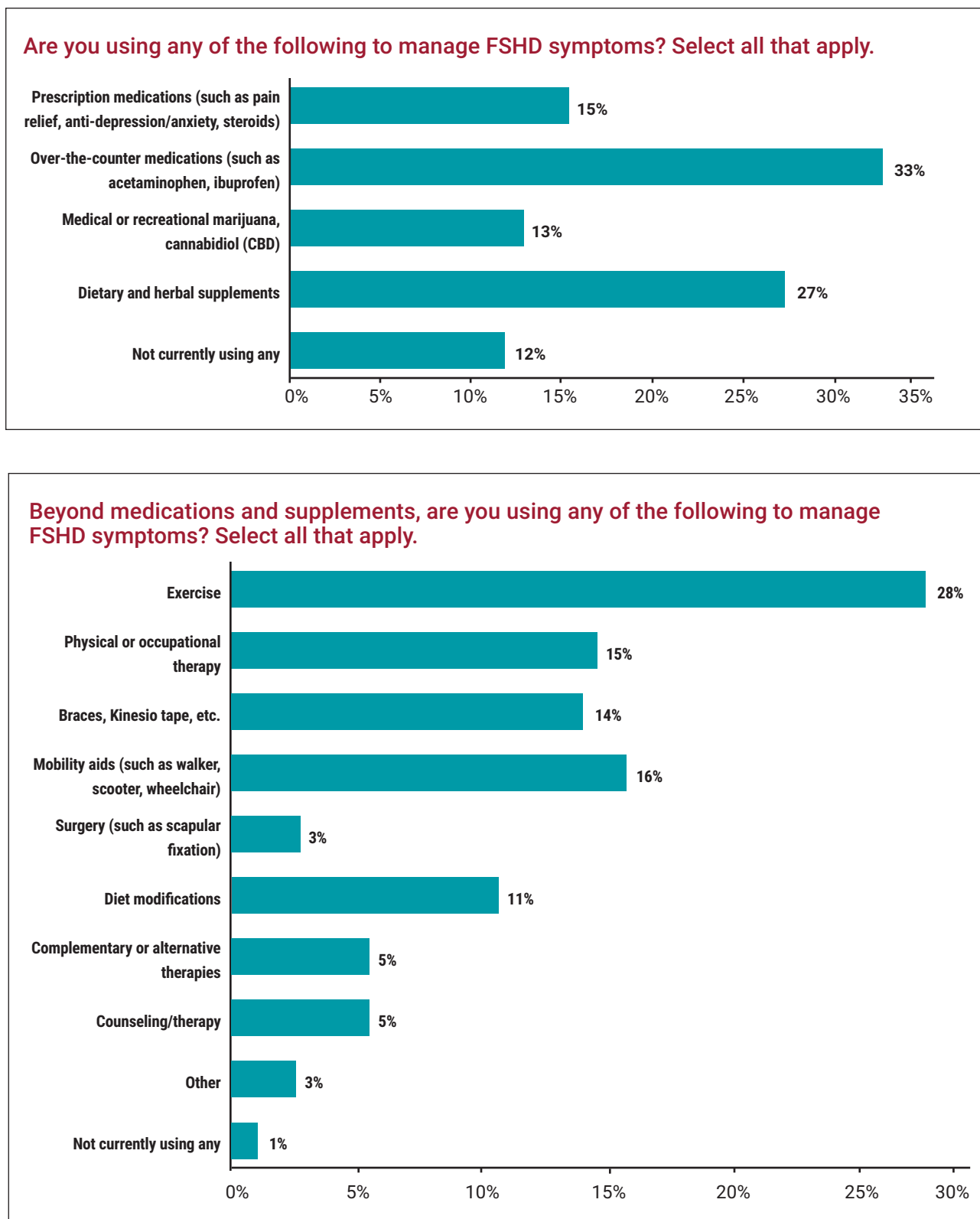
### Currently available treatments and their effectiveness

Polling results regarding currently available treatments – both pharmacological and non-pharmacological – are shown in Figure 5. Polling respondents reported that over-the-counter medications such as acetaminophen and ibuprofen (33%), and dietary and herbal supplements (27%) were their most commonly used treatments. Prescription medicines were used by 15%, and medical or recreational marijuana or cannabidiol by 13%.

Exercise was the most commonly used non-pharmacological management approach reported by polling respondents, used by 28% of respondents. Other less commonly used approaches include mobility aids such as a walker, scooter, or wheelchair (16%); physical or occupational therapy (15%); braces, Kinesio tape, etc. (14%); and diet modifications (11%).



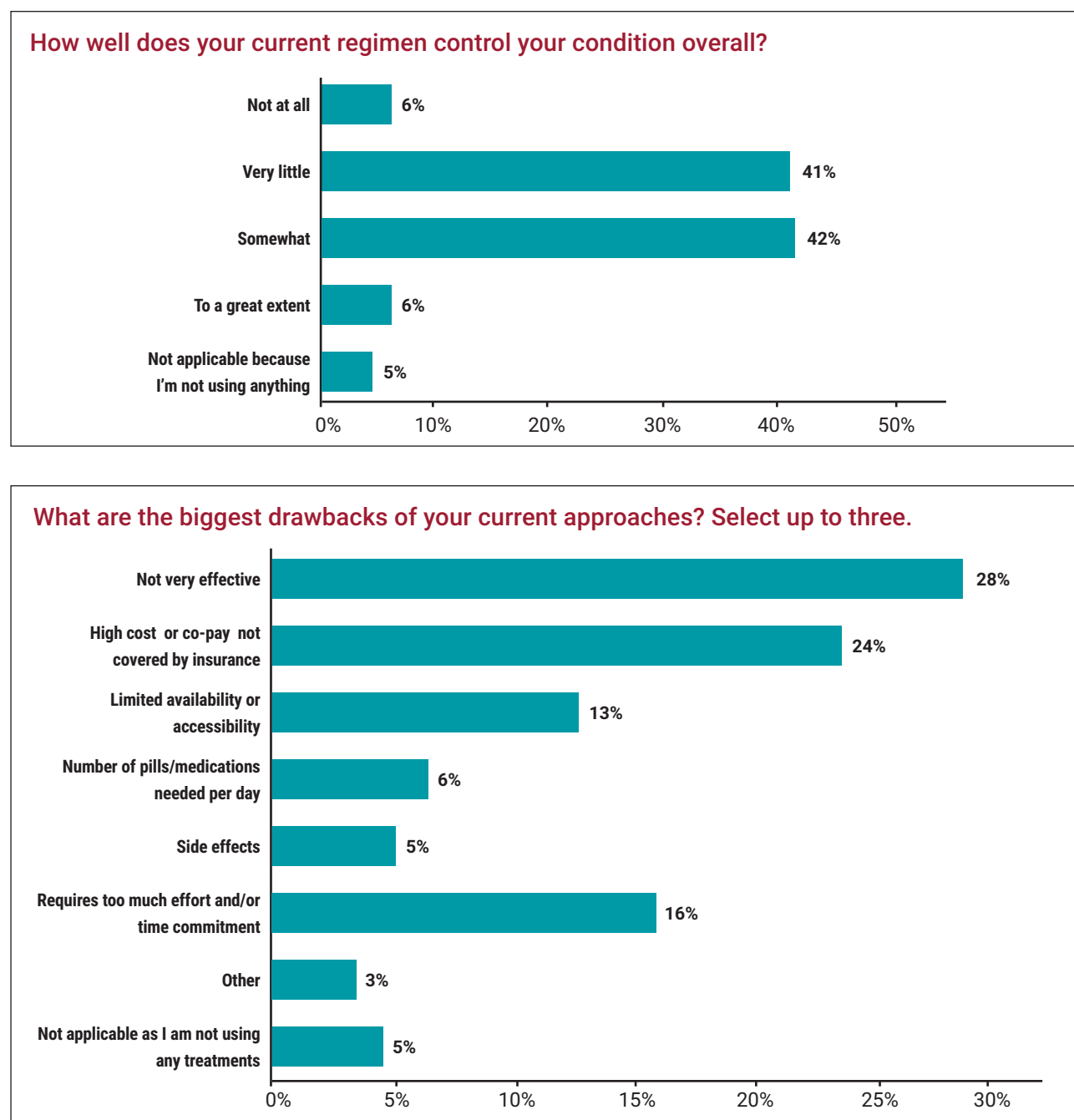
FIGURE 5. TREATMENTS COMMONLY USED BY PEOPLE WITH FSHD





Results from the polling questions (Figure 6) indicate, however, that these management approaches have limited effectiveness, controlling the condition “somewhat” (42%), “very little” (41%), or “not at all” (6%). Only 6% of respondents said the current regimen controls their condition “to a great extent.” Moreover, when asked what are the three biggest drawbacks of these current approaches, polling results indicate that in addition to not being very effective (28%), their high cost and insufficient insurance coverage (24%), effort required (16%), and limited availability or accessibility (13%) limit the ability of patients to access their benefits.

FIGURE 6. EFFECTIVENESS OF FSHD MANAGEMENT APPROACHES







The comments below from the live virtual participants describe strategies patients use to manage their disease as well as the limitations, drawbacks, and adverse effects of these various approaches. Said one 27-year-old woman, *“If I could describe my life in a single word, it would be adaptive. Every year poses a new challenge, and I creatively find a work-around to get the job done, but despite the metal armor and badass attitude I wear on the outside, I am scared underneath.”*

## OVERALL MANAGEMENT STRATEGIES FOR FSHD

A man on the Zoom panel said, *“All of my focus in life is doing everything I can with diet, exercise, and wellness, to hold out and keep the strength to be able to live independently and hold out for a cure or something that can at least stop the progression.”*

A woman wrote, *“Acupuncture has changed my fatigue level tremendously – amazing! I can do more now than I have in years, and have also been able to increase the reps of my PT exercises; belly dance is fun and has helped my core strength; meditation helps me feel focused and calm and positive.”*

The mother of a seven-year-old girl with early-onset FSHD wrote, *“She gets wonderful support and special education from her school, which describes her as having moderate to severe special needs, and gives her speech therapy, occupational therapy, physical therapy, adaptive PE, and specialized academic instruction.”*

A woman wrote, *“One positive treatment is reaching out and helping others whenever possible. Giving a friend a call, sending a text or email, paying kindness forward, being involved with meaningful events are small examples of living a meaningful life while coping with this disease.”*

## PHARMACOLOGICAL TREATMENTS

A 27-year-old woman said she started taking prednisone at about age 10: *“Although clinically this hasn’t been proven to be effective, it reduced my number of falls and the severity of my fatigue. I was started on a low dose and would take it for about six to eight months; then I’d be weaned off for a few months. Then I’d be put back on it with a slight dose increase. The prednisone suppressed my immune system, putting me at risk for respiratory infections. I would get pneumonia sometimes twice a year.... I’d also experienced rapid weight gain and trouble sleeping. I often felt highly energized with difficulty settling down or focusing. Once I was informed of the potential long-term side effects of prednisone coupled with many visits to the emergency room for pneumonia, I decided the benefits weren’t worth the risk anymore, so I’ve stopped taking the medication.”*



A 45-year-old man said for many years he took the calcium channel blocker diltiazem. *"My folks ... did a lot of research back in the late '90s after I was diagnosed that showed that blocking calcium in the muscles would prove to be beneficial. And there was since a study done ... that showed that it was not effective. I did see some improvement.... I certainly felt more fluidity in my gait, more confidence in my movements.... I started to have other side effects from the medication after a number of years – intestinal issues and lightheadedness – so I stopped."*

One woman said, *"Cymbalta helps with pain."*

A man wrote, *"I've had excellent results with a specialized mesenchymal stem cell treatment. It's expensive but made a massive difference with fatigue and energy."*

A woman wrote, *"The only treatment that helps is the antidepression drugs, which don't help the symptoms but it helps with attitude and thoughts. Without it, people really don't want to be around me."*

Another woman wrote, *"I have taken many brands of depression medications, and the medications Lyrica, gabapentin, and tramadol for pain. All of the medications resulted in weight gains of 25 pounds or more. As my FSHD progresses I am even more unable to exercise to lose this added weight. The added weight makes it only harder to stand up, and to sit comfortably, and is not healthy in regards to heart health and diabetes risk."*

## SUPPLEMENTS

A 60-year-old man said, *"I take a daily cocktail of supplements: selenium, zinc, magnesium oxide, CoQ10. I feel it helps me maintain the muscle strength, but there's no real evidence of that, and there's no studies that are going to prove that it's effective, and they are expensive."*

A 27-year-old woman said she uses cannabidiol (CBD): *"I take it orally and it has really helped with inflammation. Since I also use a wheelchair, I've noticed that, because I don't walk anymore, my legs and my ankles specifically get swollen. And when I do not take the CBD oil every day, I notice that I get pain in my ankles and some swelling. So CBD has really helped with that."*

Other people mentioned taking creatine, selenium, zinc, and CoQ10 supplements, but did not know if they made a difference. One man wrote that nicotinamide riboside (Niagen) *"... has had a big impact over the last two years."*



## PHYSICAL THERAPY AND EXERCISE

A 60-year-old man said, *"I'm riding a stationary bike 20 to 30 minutes three or four times a week. I've also incorporated a light but moderate exercise routine which includes using some weights, loop bands, and then finishing up with some light stretching. I have experienced less fatigue and a positive effect on my quality of sleep, but I don't feel that my muscle strength has changed any due to the exercise over the last one-and-a-half years."*

Another man said, *"I'm very consistent with a daily exercise plan; consistency is slowly working to make me a bit stronger and more independent. I think, to me, the biggest thing is to have a calm mind and be able to focus on being positive, and continuing to move forward."*

A woman on the Zoom panel said, *"The best thing that worked treatment-wise is getting in the pool with my physical therapist."*

A 53-year-old man said, *"The thing that sort of keeps me going is exercise, and it's made a significant difference.... I was a bicycle racer for a long time until my mid-30s when my right leg and then left leg started to go, but I still ride regularly. I think it's a great exercise, like swimming. It's non-weight-bearing, which I think is important."*

A man wrote, *"Swimming has helped me. I'm using a power wheelchair full time, but I am still able to swim laps on my back. Swimming has given me a great degree of independence. This is one of the very few activities I still am able to do on my own."*

Another man wrote, *"I have been practicing kung fu and qigong to help fight back against FSHD. When I first saw the neurologist at Johns Hopkins, after examining me, his comment was, 'Whatever you are doing, don't stop.' These practices may not be doing a lot physically, but it is a mental battle that I am fighting, too."*

The mother of a girl with infantile FSHD noted the high costs associated with physical therapy: *"She has been going to physical therapy now two times a week for three years.... The cost, however, is \$450 a week per session, \$900 total. We also invested in a stationary bike that she rides four to five times a week for half an hour.... We had to buy the high-end bike to fit her little body properly, which was a cost of \$2,500."*

A 50-year-old man said he has been riding a spin bike for 20 years even as his disease has continued to progress. *"As you get older, it's really hard to see the benefits. So you're banging away and working as hard as you can, but at the same time you feel like you're pushing a boulder up a hill.... You're not making any progress. It's a real test of one's will to continue to put the time in, not knowing if it's really doing anything or not."*



Several people mentioned that with gyms closed during the COVID-19 pandemic, they feel that their disease has progressed, and they have been unable to do what is required to maintain function, mobility, and strength.

## ALTERNATIVE THERAPIES

A woman on the Zoom panel said, *"I've been doing almost daily yoga for about 20 years or so and a meditation practice.... I've been to PT enough over the years to know what they're going to tell me to do, and now I can do it in my yoga practice. Keeping my mind focused and on the positives [through meditation] is essential to get through phases that are rougher than others as I see my disease progressing and there are more limitations physically; I have to keep myself mentally strong."*

A man from Brazil said he has tried anabolic steroids, anti-inflammatories, cortisone, vitamins, and CBD, but the only treatments that made a difference were Ayurvedic treatment and yoga.

A woman wrote, *"Massage therapy can be a great benefit for pain relief, increased circulation, improvement of attitude, and reducing contractures."*

Another woman wrote, *"Rolfing helps my two sides be better balanced. Restorative yoga helps maintain movement."*

A 27-year-old woman said, *"I do a lot of sound bath meditation therapies and stuff to really help to build that mental fortitude.... The meditation and the sound baths really help with just managing the stress, the anxiety, and also just the depression that comes with FSHD."*

## SURGERY

A 27-year-old woman said she had spinal fusion to correct scoliosis and prevent permanent damage: *"Having the surgery was not an easy decision. I was told that after surgery it was likely that I wouldn't be able to walk anymore because my FSHD had already progressed so much.... I chose to go ahead with it. The surgery successfully corrected my spine, but I lost the ability to walk independently."*

Another 27-year-old woman had surgery to address pain: *"I had scapulothoracic fusion surgery.... It's a risky surgery because you never know how your body will react. It could cause lung problems with the wires snapping or pain from the wires digging into your skin, etc. Basically, the surgeon fuses your scapula bones to your rib cage and uses wire to hold it in place while it fuses. The recovery was about eight weeks. And then several months of physical therapy. There are many pros and cons to the surgery, but for me, the pros outweigh the cons."*



Another woman who had scapular fixation surgery added, *"It was a very difficult recovery.... It was very long, it was very painful, much more so than I thought."*

A man said, *"I had the lower lid eye surgery where they lift the lid to where my eyelids can come down to, and that's really helped to relieve the strain on my eyes, especially when I go outside, and it helps with sleeping because my eyes aren't open the whole time."*

The father of two adult children with FSHD described the struggles they had with deciding about surgery for their son: *"We really wrestled with [whether he should have] spinal surgery to correct his lordosis, which was associated with his muscle weakness.... We were concerned that he would lose his remaining ability to walk.... We waited until my son started getting some ulcers.... And once he started getting the ulcers, we knew we had to do the surgery. So it was a very difficult situation for us. He was 18 years old at the time, and he knew one of the sequelae could be that he would not be able to walk again. So he had the spinal surgery, and, of course, he was not able to walk anymore, so it was very difficult...."*

After the surgery, *"... he was able to digest better. He did not have any of the ulcers anymore. He was able to sit up straight and didn't have to constantly worry about his balance falling over while he was using the wheelchair ... he did not require personal assistance, so he was able to live on his own."*

## ASSISTIVE DEVICES

A 27-year-old woman said that at age 21, she *"bought a scooter, a car with a lift, and ramp for the house that I lived in. The scooter collected dust, though, for six months because I knew people would treat me differently. Late spring of 2016, I took it for a spin and it felt almost like running again, though new challenges presented themselves due to the progression."*

Many people mentioned the use of ankle-foot-orthoses (AFOs). For example, a 60-year-old man said, *"I'm wearing a carbon fiber ankle-foot-orthosis brace [AFO] to help with the foot drop on my left foot.... It does help reduce the fatigue when I walk long distances, and it definitely helps lessen my anxiety about tripping.... I use a cane to help with balance."*

A 53-year-old man said he had tried AFOs but found them uncomfortable and fatiguing. Instead, he uses high boots with a stiff ankle, which gives him stability and allows him to walk better.

A 47-year-old man said it took a while to adapt to using AFOs while driving, and made walking up and down stairs difficult. He continued, *"They helped me with some muscle groups, but also caused other muscles to gain more atrophy from less usage.... At first, I did well, but when I turned 40 everything seemed to change. Even when I was wearing my AFOs, I began to fall. It seemed*



*like everywhere I went there were obstacles. My doctors recommended physical and occupational therapy, and after sessions and consultations with them realized we would need to renovate our house to make it handicap accessible. Those changes included an elevator lift, accessible bathroom, doors, cabinets, and less steep stairs with special handrails and a ramp.... With the elevator lift I didn't have to climb stairs and take the risk of possibly falling, but now my legs were getting weaker.... Things would get better for a few months and then another muscle group would become weaker. I would use a cane for about year and then needed to move on to a mobility scooter and eventually a power wheelchair. Then, I needed to purchase a van with a ramp to accommodate the scooter or power wheelchair. I was happy because I could remain independently driving; however, as the months went on, I then had difficulty moving my foot from gas to brake. I had a minor accident, and after that situation I decided for the safety of my family and others on the road it was time to stop driving."*

A 35-year-old woman said, *"At 19, I got my first AFO to prevent falling and to help me walk. It was uncomfortable, hard plastic that required bulky athletic shoes, two sizes bigger than normal. Needing to wear an AFO felt like my autonomy over how I present myself in the world was being taken away, and it hurt so badly that my toes would become numb. Today, I wear a carbon fiber AFO that is more comfortable, but just as ugly. It does nothing to treat my FSHD from worsening.... I also started wearing a corset-style back brace.... Its interior, sharp metal bars slide out of place, bruising and stabbing my back and abdomen. The back brace does help me stand and walk for longer, but it does not treat my worsening FSHD."*

## **BiPAP AND COUGH-ASSIST MACHINES**

A 27-year-old woman said FSHD has made it difficult for her to eat and breathe. *"I was given a BiPAP machine to use at night. I've had the machine for about five years now and I still struggle to use it daily. I sometimes wake up in pain with an extended belly full of swallowed air, or I had to remove the mask because my nose and throat burned so much in pain from them being dry.... If I am successful at using it for more than five hours, taking it off in the morning can be jarring. It feels as though that someone has suddenly turned down all the oxygen in the room."*

The mother of a 17-year-old woman with early-onset FSHD that has left her with weak core muscles said, *"She uses a cough-assist machine to help her take a deep breath on days when she's feeling particularly tired or weak."*





## DIETARY CHANGES

A 27-year-old woman said, *"I've changed my diet and the way that I eat. I avoid foods that are hard to chew or swallow and eat smaller meals to avoid fatigue, which can sometimes lead to aspirating, and that list of foods is forever growing."*

## MANAGING PAIN

A 27-year-old woman said that when pain medications proved ineffective, her doctors recommended steroid shots. *"We felt that this treatment wasn't successful and not worth continuing. Since then, I've had trouble finding something that actually helps with pain management. I've used THC [tetrahydrocannabinol] pain cream, CBD cream, back braces/therapy, and acupuncture. Majority of those treatments really didn't do anything for me. THC cream is one of the only things that I've found that has helped with pain ... it doesn't take away the pain. It just makes it a bit more bearable."*

A 57-year-old woman described how she manages pain: *"I try to do things to try to keep moving. I think that keeps the joints going. I find that when I do a very low-key workout, my back feels a little better. The ankles hurt no matter what."*

A man phoned in to say that every three months or so he has nerve ablation: *"I have ablation of the nerves in different parts of my spine to kill the nerves off, to help with the back pain. That does help quite a bit, but it just doesn't last long enough; insurance won't let them do it as frequently as I would like. So between that, I do have needling where they do put in some ketamine or something like that, also done to relieve the back spasms."*

One woman said she had tried physical therapy to manage pain: *"I ended up feeling more pain and also less stable on my feet than before therapy. So I have given up on physical therapy and currently available medications. The only thing that offers me comfort is a heating pad that I rotate around different muscle groups on my body. It only relieves the pain while in place, but it is something that I can use as often as I like without any side effects."*

Several people also mentioned concerns about the potential for dependency or addiction to pain medications. Said one woman, *"When facing severe pain daily, we're sitting ducks for becoming drug addicts. Prescription pain meds are highly addictive, don't work for long, quit helping, or devastate your liver and kidneys."*

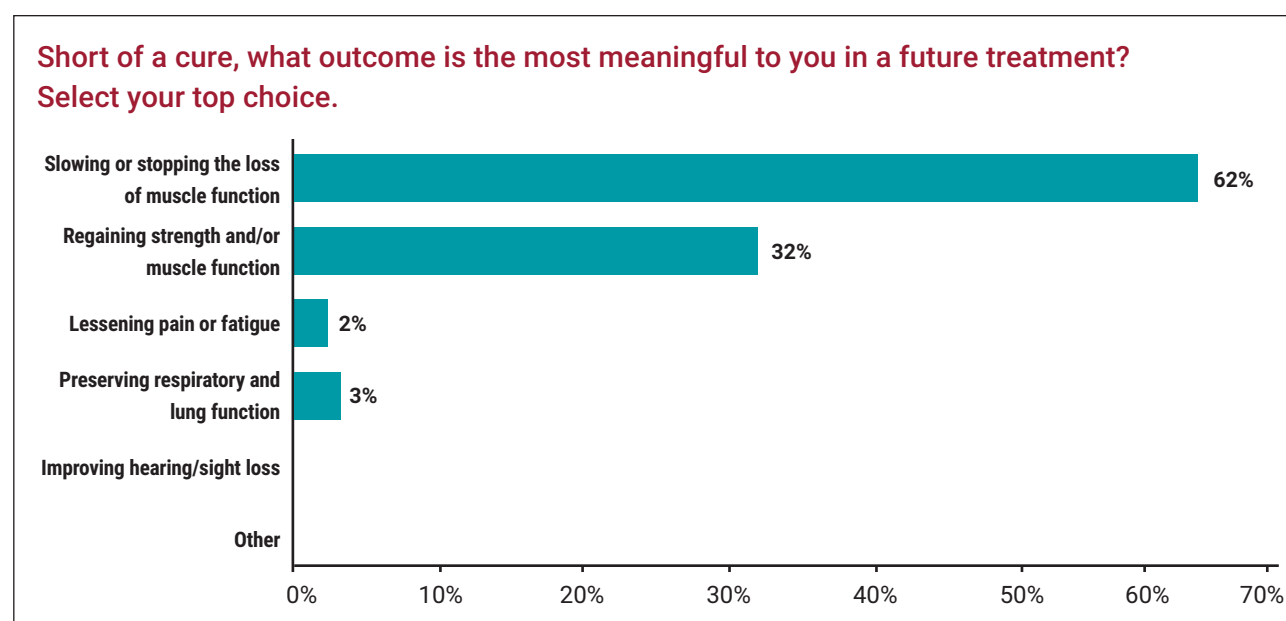




## Hopes and expectations for future treatment

Short of a cure, the majority of survey respondents indicated that slowing or halting the disease progression to stabilize their status was a highly desired outcome. Survey respondents signaled their preference for slowing or stopping the loss of muscle function (62%), followed by regaining strength and/or muscle function (32%). A common theme in the comments shared during the meeting is that people with FSHD are willing to tolerate a higher degree of uncertainty about the effectiveness of a new drug, given the dire, unmet need they currently experience.

FIGURE 7. WHAT OUTCOME OF TREATMENT IS MOST MEANINGFUL?





## STOPPING OR SLOWING DISEASE PROGRESSION

*"Treatments to halt progression and/or improve muscle strength would be life changing," wrote the mother of a 17-year-old woman with early-onset FSHD.*

*A 26-year-old woman said, "Having a significant treatment for FSHD would really be life changing.... I would like to see something that would stop progression of the disease. If I were to stop progression right now, I would still be able to walk in 10 years. I would still be able to smile, to get off the couch, to raise my arms, to hold my future baby and countless other things...."*

*A 50-year-old man said, "For those of us who are still lucky enough to be able to exercise, being able to continue to do that for hopefully the rest of our lives has just huge, not only physical benefits, but it's got a lot of also psychological benefits.... When you're able to exercise, you start to fire off these serotonin and dopamine reactions in your brain. And that really has a huge impact on attitude and behavior and mood, and being able to lift your mood to take on the battles that we all have to take on every day. I think it could pay huge dividends."*

*A 35-year-old woman said, "We need treatments that address the actual disease. Treatments need to stop patients from getting sicker and help us get better, and these treatments need to be affordable and accessible to everyone. Treatments need to help us regain strength, to not live in pain, and to be able to enjoy life's simple pleasures like picking up a child and cradling it in your arms, walking and scaling life's mountains without having to worry about the disease burden."*

*A 27-year-old woman also mentioned the importance of how a treatment is delivered: "Is it oral? Is it an injection? Do you have to go to another location to get the treatment, and what are the side effects after it? When I'm thinking about needles and injecting it, I have low dexterity in my hands. So I won't be able to really inject anything into my muscles. So for me, I think it would be beneficial to have an oral tablet."*

*Another woman added, "This disease is highly variable, and a drug that just makes slight improvements for one person might make huge improvements for another. A drug that doesn't seem that helpful for someone who is very progressed in the disease might be helpful for someone barely showing symptoms. For this disease, even something that makes a slight difference is a worthwhile treatment."*



The mother of a young daughter with FSHD observed, *“Everything I have read about FSHD proves it is unpredictable and variable. I can attest to the fact that one day [she] was fine and the next her shoulder was slouched and had lost muscle ... overnight! If there is a drug candidate on your table that does no harm and can potentially help, we NEED it. Time is not on our side. My daughter is not disabled yet. She is 11.”*

Regarding herself and two affected family members, a woman wrote, *“We want a treatment that will stop the progression of this horrible disease and are willing to risk a treatment that is safe and promising, even if it has not yet been proven effective.”*



## 7. Incorporating patient input into a benefit-risk framework for FSHD

In 2013, the FDA published a draft implementation plan for a structured approach to benefit-risk assessment in drug regulatory decision making and updated this plan in 2018. The current plan reflects a requirement of the 21st Century Cures Act that the agency issue guidance on how patient experience data will be incorporated into the structured benefit-risk assessment framework to inform regulatory decision making. This framework calls for assessing four decision factors: Analysis of Condition, Current Treatment Options, Benefit, and Risk and Risk Management. When completed for a specific product, it summarizes each decision factor and explains the FDA's rationale for its regulatory decision. The benefit-risk framework is important for both regulatory and treatment decisions.

The PFDD process allows patients' voices to inform the development of a benefit-risk framework for use in the evaluation of new treatments. People living with the disease have a unique perspective on the dimensions that are most important and critical to regulatory decision making, the unmet medical needs of others with their condition, and the benefit-risk tradeoffs that may be acceptable across the continuum of the disease. Their input thus should guide therapeutic development to ensure that treatments have clinical meaningfulness and address aspects of disease that are most critical to people living with the disease.



By reflecting the perspective of people with FSHD, drug developers will be better able to design clinical trials with a high chance of success. Defining the benefit expectations and risk tolerance for varying treatment options will also enable better characterization of tradeoff decisions faced by patients, families, and healthcare providers, as well as regulators.

The input provided by people with FSHD and their caregivers is summarized here in this sample framework (Table 1) to provide an understanding of the benefit-risk aspects for these decision factors. This sample framework is likely to evolve over time and could be incorporated into a benefit-risk assessment framework for a drug under review.

This Externally-Led PFDD meeting demonstrates that people with FSHD are highly engaged and enthusiastic about working with the FDA and drug developers to advance better treatments for this debilitating disease.

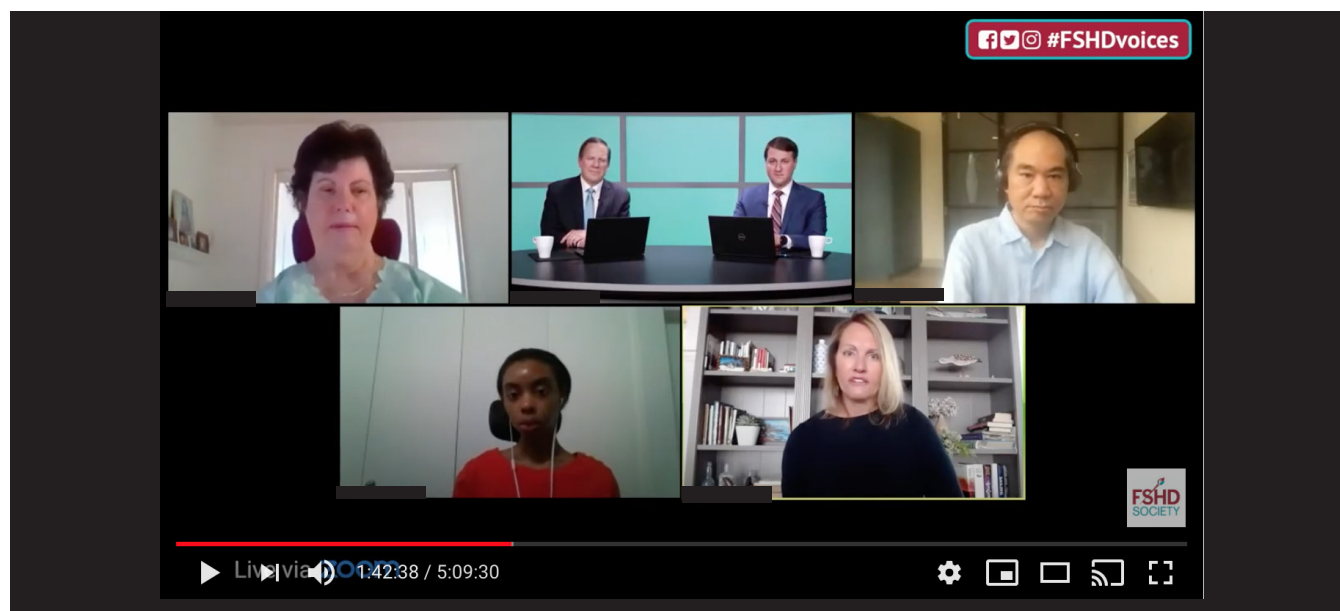
**TABLE 1. SAMPLE BENEFIT-RISK DIMENSIONS FOR FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY**

DECISION FACTOR	EVIDENCE AND UNCERTAINTIES	CONCLUSIONS AND REASONS
Analysis of condition	<p>Facioscapulohumeral muscular dystrophy (FSHD) is a progressive genetic disease usually caused by an autosomal dominant mutation. Muscles of the face, shoulder blades, upper arms, core, and lower limbs are the most severely affected. Symptoms of the disease typically are diagnosed in adolescence or early adulthood, although severe and rapidly progressing early-onset forms may be evident in infancy or early childhood.</p> <p>Presentation of disease is highly variable and heterogeneous, with a progression that is unpredictable over the lifespan. Muscle atrophy and weakness often result in mobility problems, difficulty using arms and hands, fatigue, pain, and increased falls. Pulmonary insufficiency affects a subset of patients and can contribute to fatigue, increased hospitalizations, and premature death. Rarely, people with FSHD may also have cognitive, sensory, and emotional impairments.</p>	<p>FSHD is a progressive form of muscular dystrophy that may result in severe physical disability, loss of function and independence, reduced quality of life, shortened lifespan, and substantial burden for patients and families. The severity, insidious nature, and unpredictability of FSHD take a high physical and emotional toll on patients and families, producing a high, unmet need for more therapeutic options.</p>



DECISION FACTOR	EVIDENCE AND UNCERTAINTIES	CONCLUSIONS AND REASONS
Impact on activities of daily living and quality of life	People most frequently mentioned walking and participating in sports or hobbies as significant aspects of FSHD, reflecting the impact of impaired mobility, weakness, and fatigue. Other highly impacted activities include going out, socializing, traveling, and performing household tasks.	The progression of symptoms leads to loss of mobility, impaired quality of life, and loss of independence.
Current treatment options	<p>There is no effective treatment or cure for FSHD. The life-long, progressive nature of FSHD forces patients to continually adapt to changes in function. The management of ever-worsening symptoms is time consuming, costly, and disruptive. Patients report fear, anxiety, and stress when considering the unpredictable but unrelenting progression of symptoms.</p> <p>Current treatments are primarily symptomatic and may be associated with limited effectiveness, significant adverse effects, and high costs.</p>	Drug treatments are urgently needed to slow or stop disease progression. Reversing the muscle loss associated with FSHD would also be highly desirable. Clinical outcome measures that assess aspects of the disease that are meaningful to patients are being developed and validated to accelerate the development of new therapies.
Prospects for future treatments	There is a broad scientific consensus that FSHD is initiated by the ectopic expression of <i>DUX4</i> , and on the tractability of this disease mechanism as a therapeutic target. Disease-modifying treatments are being investigated and have shown some positive impact on disease mechanisms.	<p>Slowing down or stopping disease progression would benefit patients by helping stabilize them in their current state. Therapies that reverse muscle loss and build pre-existing function would also be highly desirable.</p> <p>Given the urgent unmet medical need and the trajectory of this condition if not treated, great effort should be given to develop and approve proven treatments; given the heterogeneity of the disease, care should be taken to evaluate the potential to impact one or more of the important factors in FSHD rather than trying to address the entire spectrum of the disease, which could take many more years to develop.</p>





## 8. Conclusions

The FSHD EL-PFDD meeting was very effective in elucidating the experiences and preferences of patients and caregivers. This was due to the high degree of engagement with the PFDD process by the FSHD community, which included giving input through various pre-meeting surveys, participating in the live meeting, and providing additional post-meeting statements. All of these factors contributed to generating this comprehensive set of patient experience data.

**Key take-aways from the FSHD PFDD meeting can be summarized as follows:**

- FSHD is a serious and debilitating disease that is highly heterogeneous in terms of the types of symptoms experienced, the severity of those symptoms, and the speed of progression.
- Muscle weakness from FSHD results in significant disability and pain, with loss of mobility, ability to perform tasks of daily living, caring for family members, social interactions, and independence.
- Early-onset or infantile FSHD may be especially severe and more rapidly progressive, and may involve physiological systems not typically associated with the disease.
- Loss of independence is feared by many people with FSHD as well as by the parents of children with this disease.
- The knowledge that symptoms will worsen over time imposes additional psychological, financial, and social burdens on both patients and their families.
- There are no approved medicines for the treatment of FSHD. Current treatments are not very effective and may be associated with severe side effects.
- There is an urgent need for treatments that slow or stop progression of FSHD.





## APPENDIX I: Testimony from live webcast on June 29

### Symptoms and impact/burden

#### SYMPTOM: UPPER EXTREMITY WEAKNESS

**Pencie:** “I can’t get my arms up to wash my hair. I’ve recently gone to the salon to have my hair washed. I almost can’t get a fork to my mouth because of my arm limitations.”

**Julie:** “A recent finding is that Noah is unable to raise his arms, to comb his hair unless he uses one arm to support the elbow of the other arm.”

**Jeff J:** “As the disease progressed through my early to mid-30s, my legs and arms grew very weak. I wasn’t able to pick up my kids out of their crib or throw them in the air like many of my friends did with their kids. I had to sit down as my wife handed them to me just like she did with their great-grandmother who was fragile and weak... Other falls have resulted in a pulled groin, bloody face, torn meniscus, bruised hip, bruised elbow, broken rib, broken finger, scuffed up knees, black eye, and intense headaches from hitting my head on the ground. Now at age 50 walking is limited to a few steps inside my house, and at this point it’s easier to list the muscles I still have versus the ones I’ve lost. I still have decent strength in my forearms, left calf, and my face; otherwise, every other muscle is a shell of what it once was. This has made the simplest tasks difficult if not impossible. Forget doing laundry, the clothes are too heavy. Forget making the bed, I’ll end up on the ground. Forget loading the dishwasher, the plates are too heavy, and forget carrying a glass of water, I don’t have the balance.”

**Manuel** (regarding his older daughter): “In her kitchen, she can barely reach into the first level of shelves. If she drops something, picking it up is out of the question if it is outside the range of a grabber.”

**Carden:** “I began holding my wrist with my other hand when eating.... I’ve given up writing with paper and pen because I have low dexterity.”

**Bree:** “I noticed scapular winging when I was about 13 or 14 years old. It really affected my quality of life – affecting my ability to play softball and I was extremely self conscious about my shoulders and continue to be to this day. I tried physical therapy, acupuncture, the chiropractor, as well as some other remedies. I had no idea that FSHD even existed until last year when I was 30. I was diagnosed two days after my 30th birthday. While a relief to know what has been going on with my body for over half of my life, it is also difficult to know that the disease could progress



more and to not know when or how much function/mobility I will lose. This is so difficult to deal with personally, but also to be able to explain to friends and family. Also, I wish more doctors and medical professionals knew about this disease and there could be more research done to expand and understand treatments and how they work.”

**Priscilla**, age 38 and recently diagnosed: “Now I have lost so much functionality that I cannot care for my kids without support. I cannot carry my kids, lift them out of the crib, and my right arm has atrophied so quickly that I can only type or use my phone for limited periods of time... Doing my hair can take up most of my energy for the day or even my makeup, so I do not go out very often because of the effort and my self-esteem has suffered.”

### SYMPTOM: FACIAL WEAKNESS

**Carden**: “Chewing with my mouth closed became impossible due to the facial weakness... my smile and facial weakness, expressions weaken with every year.”

**Ellen**: “In my career I had, I was fortunate to have a fairly long career, giving presentations used to be something that were effortless and almost spontaneous, and now it’s something that is greatly complicated by the continued loss of my facial muscles. A speech or a brief presentation, now, must be carefully scripted beforehand. Each word I use must be weighed. It, or will it be difficult to pronounce? Is there another word that would convey the same emotion in a meeting that might be easier for me to use? When I’m giving a presentation, I can see the faces in the audience, as they try to understand me.”

### SYMPTOM: LOWER EXTREMITY WEAKNESS

**Jeff J**: “Playing golf, a game I loved was no longer an option because my quads were not strong enough to support my weight. Then came the falls which are often unannounced and terrifying. A strong breeze can knock me over, uneven pavement will cause me to trip and fall, being bumped into will knock me off balance and I’ll go down, or my knees will just unexpectedly buckle which will put me on my back.”

**Ellen**: “I miss the role of being an active member of my community, whether it’s in a volunteer position for a school, or serving on committees, or serving on boards and the like, and one of my issues is that I can no longer stand at a podium.”

**Cindy**: “In my freshman year in high school I was a cheerleader. By my sophomore year, I couldn’t stand up from the ground by myself... And there’s the day-to-day things that people take for



granted... I work on computers, but then just putting the hands out to type and to do keystrokes can get exhausting, and it's like, 'Oh, great, what's the shortest way I can convey what I need to say without having to type an extensive amount of time?'"

**Kate**, a 26-year-old woman from the UK, was diagnosed at age 13: "My parents were inconsolable, and I told them not to worry. As long as I could still ski, I'd be able to handle things." She was 13 years old at the time, but at 17, she skied for the last time. And today she says she struggles to get upstairs. "I walk with significant gait and trip a lot because of foot drop and I can no longer get up from the floor when I fall. And recently on a windy day, I fell over and had to crawl for 30 meters to find a step where I could hoist myself up. It was degrading and humiliating. It's devastating to have lost so much at age 26."

## SYMPTOM: PAIN

Many patients reported pain as the most severe and impactful symptom.

**Jeff B**, a 58-year-old man with FSHD1: "I'm in constant pain from all the wear and tear put on my body from having to compensate for the lack of muscle, not to mention all the falls. Instead of living my golden years, it's the broken years."

**Jeff J**: "Nowadays pain is a constant companion, my lower back hurts from sitting so much and the extreme hyperextension in my left leg results in knee pain after walking only a handful of steps. Being able to stand stationary relieves my back pain but eventually my right leg goes numb and I need to sit down. Finding comfort is an ongoing battle."

**Carden**, a 27-year-old woman with FSHD resulting from a spontaneous, de novo mutation: "[At age 14] my typical afternoon three mile run felt off. I couldn't propel my legs forward like they used to and a mile in I collapsed on the cul-de-sac crying, at the time, my favorite thing stripped out from under me. I gave it another year and a half and 10th grade flag football was the last time I ran.... I would leave class early because I had to use the 'bathroom' since I couldn't walk across campus before the next bell ring.... Stairs grew to be more of a challenge, and I avoided attending school rallies and football games.... First, it was grabbing for a hand rail, then it turned into using my right leg only to ascend stairs one step at a time. Falling down stairs became a weekly thing. My friends were talking about kissing boys and I was worried about not tripping and falling on the graduation stage steps. My gait and lordosis became more pronounced and I ignored the mean comments everywhere that I went. Sophomore year of college, I fractured my skull after tripping due to the foot drop. Eleven days in the hospital and a tender spot on the back of my head for the rest of my life."



**Diane**, a 57-year-old woman with FSHD diagnosed at age 18, said that in the last three or four years: “The pain has really become so bad for me, my ankles, my lower back, my neck.... As soon as I get up in the morning, I start walking for a half an hour. I have to sit down my back hurts, my ankles hurt and that just puts a damper on the rest of the day.”

**Cindy**: “There’s the spine pain; because of the curvature of the spine, I get the lower back pain constantly.”

**Susan**: “My son no longer has the muscle strength to hold up his head easily. He has daily neck and back pain due to this difficulty. He has a little curved bar behind his head on his new wheelchair. I cannot see that this is going to help much. For instance, it sure won’t stop his head from flopping forward. In any case, he takes lots of painkillers, yet the pain is severe and daily. Sometimes the pain is so bad that he vomits. He is naturally tempted to spend the day in a recliner, in a reclined position where his head is continually supported by the back of the chair. This inactivity will, of course, lead to more muscle loss in other parts of his body, but it does lessen the pain.”

## SYMPTOM: FATIGUE

Many people with mobility problems report frequent falls; however, falls may also be associated with fatigue.

**Terry**: “Due to the lack of treatment, accurate prognosis for FSHD, when my job relocated, I felt I couldn’t relocate. The new location would require a much longer commute, and I didn’t know whether I would be able to manage my fatigue, or whether it would get much worse and have any life outside of work. So I’ve gone from a well-paying, fulfilling job to living on my savings until I can retire in a few years.”

**India**: “When I was walking [I was] exhausted from walking so much and then being in a car and my back hurting and things like that. And so it’s difficult in a different way now because now my arms are so severely impacted. So that has a different impact on how I’m able to care for myself. So the pain from walking in my back and my legs doesn’t exist anymore, but now my shoulders and my arms hurt, or just now it’s the breathing issues that I have make it difficult because now I’m exhausted just from sitting up all day.”

**Pencie**, a 67-year-old woman with symptoms since her 30s but not diagnosed until 2019: “When I look at a pineapple on the counter ... I look at it and I think, ‘I don’t have the energy to cut up a pineapple.’ It’s very emotional, especially when you’ve been a doer, like I have.”



## SYMPTOM: RESPIRATORY PROBLEMS

**Daniel:** “Respiratory issues such as sleep disordered breathing, insufficiency and acute respiratory failure are of issue in FSHD patients. I’ve experienced both long term chronic progressive issues as well as acute respiratory failure due to CO2 retention. I now use a ventilator. Without doubt I am convinced that this is an unmet and misunderstood need in measuring disease progression in clinical settings and trials endpoints. Measuring tcCO2, etCo2, blood gas and metabolic measures and correcting for insufficiency would be helpful. Improving pulmonary function is important to me and I would think one of the most accurate and sensitive measures for drug trials.”

**Joseph:** “My body is effectively completely paralyzed due to FSHD and I am fully ventilator-dependent, but one of the most debilitating aspects to me of my own muscle deterioration is having lost my speaking voice. I cannot phone in to offer my comments because my voice has become just too weak and garbled. My mind remains active and vibrant -- I have a doctoral degree in mathematics – yet I can no longer express myself verbally in most situations. I cannot mime or point to compensate for my absent voice nor even write my thought on a pad of paper, because FSHD has stripped me of most voluntary muscle movement. My communication difficulties have even put my life in danger. I have been in the hospital for reasons unrelated to FSHD when my ventilator has accidentally become disconnected yet I was unable to communicate my difficulty breathing. I have had hospital nurses ignore my attempts to speak to them and turn my body into positions that again compromised my breathing.”

## PROGRESSION

**Ian,** a 54-year-old man who was diagnosed at age 17, thought for a long time that he had a milder and slower progressing form of FSHD1 : “At 50 years old, it was like a light switch that was flipped, the increased pain in my legs. Now it’s transitioned into my sleep and it’s affecting my sleep. I am losing my ability to walk. I’m losing my ability to play music. I’m a musician.”

**Jack,** 60 years old, diagnosed with FSHD1 at age 32: “The rate of progression of this disease for me, over the last three years, has been just downright frightening.”

A woman talking about her 11-year-old daughter diagnosed with early-onset FSHD at age eight: “... can barely run anymore. She can’t life both arms overhead. She struggles to walk up the stairs. One day she was fine and the next her shoulder was slouched and had lost muscle ... overnight!”



## IMPACT: WORKING

**Cindy:** “The neurologist that diagnosed me was pretty blunt. He said, ‘Change your major, get a desk job. Don’t overuse the muscles and think twice about having kids...’ I was a costume designer and ... that meant that I was on my feet, 10 to 13 hours a day, six days a week, and the day off that I had, I spent recuperating where others were off partying, doing fun things. I was just pretty much watching TV, trying to recuperate, getting ready for the next week.” Cindy was offered a job that came with no health insurance, so she turned it down and accepted another job: “it was on the second floor of a building without elevator access.... I had braces ... they had a couple of railings put in, but that was the only modification I needed. The differences really didn’t get in the way of my job until about 2010. And by that time, every morning, I just dreaded in the morning. I’d get up, and I think about, ‘Am I going to be able to get up the stairs? What’s that going to feel like?’”

## IMPACT: WALKING/MOBILITY

**Jeff B,** a 58-year-old man: “While you are walking you’re always looking on the ground for anything that might throw off your balance, sticks, pebbles, cracks, anything uneven. When you walk into a room, you have to survey all your surroundings and strategically plan where you can sit, usually by a desk so you can push off with one muscle, pull with another to stand up, or if you fall how you will get back up. When I hit my forties, that’s when FSHD started taking control of my life by taking away from me. First couple of major things that I lost was the ability to climb stairs and ladders and that was a vital part of the work I did.... I went from making \$70,000 a year down to \$18,000. All the things that made me happy, I was no longer able to do. For example, I went to a sporting event, had to use the restroom. I slipped, fell, everyone just walked past while making remarks that they thought I was drunk. I haven’t gone to a game since.”

**Manuel,** who himself has a mild form of FSHD, described the effects on his two more severely affected daughters: The older daughter (now about 35) has “severe difficulty walking, and even for short distances she needs a wheelchair.... She can barely go up steps, and she has long been unable to visit the second floor of our house. She has great difficulty sitting or getting up from a chair without falling.... If she falls, she must crawl to a bed or other furniture to pull herself up.”

**David,** 44 years old: “I’ve lost a lot of function over the past decade, fallen many times, broken my nose at least five times.”



## IMPACT: DAILY ACTIVITIES

**Jeff B:** “When I go to the store or to a doctor’s office, if the wind is blowing my body will freeze up because of the fear of falling and not being able to get back up, that is so embarrassing. I am at the point now, if it’s dark, snowing or even raining, I won’t leave the house. I have to pay someone to do any and all maintenance on everything. I no longer feel like a man, so when it comes to quality of life, I have to say I have very little.”

## IMPACT: PARTICIPATING IN SPORTS OR HOBBIES

**Julie,** the mother of a 14-year-old boy, described how at age 10, he was unable to do a sit-up. After talking to their pediatrician, they realized that he had other FSHD symptoms and had for years: eyelid open while sleeping, unable to whistle or blow up a balloon, small flat smile, winged scapula. “Although he had played sports since he was five years old, his endurance was decreasing and he wasn’t able to keep up with his friends. By age 11 he chose not to sign up for basketball or baseball. Since then he joined Boy Scouts and middle school band, but was unable to pass the swim test at Boy Scout camp, which deprived him of some swimming activities. He cannot walk with a heavy backpack for an overnight camping trip, and it is a significant challenge for him to go on a five mile hike with his fellow scouts. As he is unable to purse his lips, he was limited on the instruments he could select to play in band. However, he determined that he could play percussion and he has taken a liking to this new hobby.”

**Jeff J:** “I liked to play sports, work up a sweat, go for a hike. Now most of the things I’m passionate about have been taken from me, now I get around in a scooter which has been a major adjustment both physically and mentally.”

**Manuel’s** younger daughter was “forced to give up her avid jogging and that she now has almost completely given up her equally avid urban cycling because of growing weakness in her legs.”

**Sam,** a 14-year-old, who as a younger child was very active and played soccer and baseball: “I can’t really play any sports right now.... I can’t really walk anymore [since fifth grade].”

## IMPACT: GOING OUT, SOCIALIZING, TRAVELING

**Julie:** “Over the last four years, we have chosen to modify our family activities and schedules to ensure Noah is able to participate.”





**Cindy**, diagnosed at age 15, now age 50: “I have not walked or stood since I was in my mid thirties. Can’t do it. It’s impossible. So to go out and be in the community and society, I have to plan everything. I don’t drive anymore, so, okay. Is there a bus or transportation, plan that, Oh, there’s space? Like I go meet other people who have wheelchairs, does the restaurant have enough room to accommodate, is the bathroom accessible? ... I have to define and plan ahead for every situation, even going to the grocery store. All those little things.”

**India**, a 27-year-old woman diagnosed at age six: “I’m currently in this stage where going out is important, being a young adult, and you have to weigh what’s important and balance things. So if you’re too exhausted from trying to take care of yourself throughout the entire day, when your friends want to go out at a bar at night or go hang out, that may not be something that you can do, because you’re too exhausted. So unfortunately, sometimes I have to say, I’m really not up to it. And even traveling or being on a plane or in a car for an extended period of time, maybe it’s just being too exhausted from sitting up the entire time, or just the pain of being in the car, and then just personal hygiene.”

**Justin**, a 29-year-old man, developed early onset FSHD at age four and was permanently wheelchair bound by middle school: He said it was bad enough that his arm muscles were deteriorating, which made him lose independence, but it affected his social life in college and beyond as well. Most dorms, houses, restaurants are not wheelchair accessible and don’t have accessible bathrooms. “And now I need an aide to get me out of bed.”

## IMPACT: CARING FOR ONESELF

**Carden**: “Age 22 was the last time I used a regular stall bathroom because I couldn’t stand without grab bars. Twenty-three was the last time I drove because I couldn’t pick my foot up off the pedals. At 24 I purchased a power wheelchair because the scooter wasn’t sturdy enough for me. Age 25 was the last time I stood to take a shower because I fell often. And 26 was the last time I walked and also the last time I stood on my own.”

**Amy**: “The activities of daily living, with pain and an inability to raise your arms, is indescribable. Imagine if you will, if you were tied up by a burglar and they took a strap and they wrapped it around your entire upper body and both arms and your trunk, and you could not lift your arms more than six to maybe 12 inches, and then you want to scratch the top of your head, or you want to brush your teeth or you want to reach a shelf. And the effort that you have to exert ... in order to even get a modicum of movement towards your upper body, towards grooming, brushing teeth, dressing, putting on a shirt, showering, is so exhausting, that that’s where the fatigue and the mental challenge comes in.”



**Cindy:** “Just to be able to put on a shirt in the morning, I have to place my elbows on a desk or counter top to put on a shirt. To wash, I have to put an elbow on the wall to get to wash the hair, to get the hands up. Different adjustments, having in order to eat, raise one hand that’s got the utensil in it with the other one, to lift it up to the mouth.”

## IMPACT ON FAMILY LIFE

A 58-year-old man: “I made a decision not to have children after watching all of the troubles and heartache my mom and grandfather went through in their battle with FSHD... Nowadays, I feel not having children was a bad decision, I could definitely use the help from them now. Most of all, I missed out on so very much. Everyone should be able to have the experience of having children and grandchildren, even though I am unable to lift and hug a child.” He added: “I lived by myself for 10 years before I was introduced to the lady I live with now. I am so worried that I will lose her because I am unable to make love to her because of the disability.”

A 54-year-old man described similar losses: “Six years ago today, I was blessed with my very first grandchild. Recently we were blessed with two baby boys that I can’t hold now. My first grandchild I was able to pick up and hold. My two boys, I have to wait until they are old enough to walk before I can take them anywhere or be really involved in their lives like I’d like to be.”

A 61-year-old man diagnosed at age 34 described how he was excluded from a family gathering: “I guess the problem is that accommodating for somebody with a physical disability, i.e., a power wheelchair, using a power wheelchair, just might be a bit too difficult for some people... if I were able to go and visit, I would not be able to use the bathroom... Why can’t I go visit my friends? Why can’t I go visit my family? Why can’t I participate in these activities? And the reason is in many cases, because you just physically cannot do that.”

A woman from Canada commented that beyond family members being unwilling or unable to make the accommodations needed, the disease impacts those family members as well: “Spouses have to watch someone that they love decline and deteriorate, and it’s not just weeks and months they have to watch this... the loss is profound. And many times family and friends cannot cope with this emotionally. It is devastating for them to watch as it is for parents to watch their own children decline in this way.”

A 67-year old woman described a scary situation while watching her granddaughter: “About two weeks ago, I was went to visit her. We were taking a walk and she took off like a rocket at two and a half years old towards the highway... And I couldn’t catch her. I can’t run anymore. Luckily, she stopped at the road. It’s just beyond devastating.”



A woman: “I was always very active, and I have a very active family. It is very hard to be the person who limits our activities. We have to pick shorter hikes, shorter camping trips, and take more breaks on family vacations, etc., because mom can’t keep up. It also takes me longer to recover after an active day, which just puts more limits on what we do. What’s hard is knowing that this will only get worse. Eventually, our family won’t just be shortening a family hike, but maybe not be going at all, or maybe leaving mom at home.”

## IMPACT: EMOTIONAL

**Jeff J:** “The physical and mental pain is accelerating and my optimistic nature is being tested like never before.”

**Manuel** (regarding his older daughter): “Despite her enormous emotional strength, she sometimes has panic attacks and struggles with loneliness and depression.”

Manuel’s younger daughter has a milder form, but: “She’s very, very scared because she has seen the decline in her sister’s condition over the years. So scared in fact, that we fear she tends to put distance between herself and the rest of us. Anxiety and depression are also her constant challenges.”

**Cindy:** “When you lose something, it’s like a cyclical grieving process. It’s like losing a loved one and you go through the whole thing and then the next day you wake up and everything is good. You stabilize, and then you lose something else, and it’s just like losing another loved one.”

## FUTURE WORRIES: LOSING INDEPENDENCE

**Julie** (regarding her son): “All of these examples seem like fun activities that he can’t participate in or trivial things that he can’t do or enjoy. However, it is not too long before more meaningful activities are limited. Normal things like climbing stairs, carrying luggage, and reaching for something out of the cabinet we take for granted. These limitations are around the corner for Noah.”

**Jeff J:** “This disease is wicked and cruel in many ways, but losing my independence is probably the most frightening and helpless feeling I have ever had.”

**Manuel** (regarding his older daughter): “She’s now 36, living independently and working, with her own car and condo... We especially worry about how long she will be able to continue to live independently in her condo, or drive her car, or when and how she will have to modify them both substantially.”



## FUTURE WORRIES: LOSING MOBILITY/ABILITY TO WALK

**Meredith:** “I’ve fallen by myself many times, and I think one of the scariest things about falling, especially by yourself, is I’m scared that I’m going to hit my head, and I’m scared that someone’s going to find me unconscious. And I’m scared that, one day I’ll fall and I’ll break something, and lose even more mobility.”

## FUTURE WORRIES: DISEASE PROGRESSION

**Carden:** “I think about the last time I’ll be able to choose solid food because choking occurs. I worry about deteriorating into absolutely nothing but a bag of bones when all the muscle and fat dwindle away.”

**Stuart:** “It’s been a worry much more now because of the rapid progression that the disease took in my case... I fell leaving the office because I was fatigued after a full workday. I stumbled in the office hallway, fell awkwardly and snapped my femur in two. And I dedicated a whole year trying to get back to walking. That was not successful and I started using a wheelchair. So at age 45, I had no idea that I would all of a sudden be faced with that.”

**Colette:** “Waiting for a treatment is scary. I am fearful of what else I will lose while I wait. Will FSHD take mobility from my hands? Will breathing become more difficult? What else will it take away from my family members who are sick?”

## FUTURE WORRIES: STRESS OF NOT KNOWING HOW DISEASE WILL PROGRESS

**Julie:** At the age of 14 he is learning how to accommodate for his muscle weakness. I worry about the changes and modifications he will have to make throughout his high school years, college years, and adult life.”

**Manuel:** “Any conversation about a permanent wheelchair or a scooter is extremely painful, both because of the fear it creates among all of us and because nobody knows what the progression of the disease might or might not be.”

**Tim:** “You don’t know what the next step will bring, for some, or for others, in my case, the next week, or the next month or even the year, what will I look like? I was diagnosed four years ago, at 40, and it felt like someone started a stopwatch as soon as I heard I had so little amount of time to physically enjoy the things I was enjoying... the scary thing is, you don’t know how much time you have left. And I understand that that is life in general, but for FSHD it seems like it’s awful quick, and the time you thought you might have, you don’t. It’s abbreviated, because it’s so unpredictable.”



Ian, 54 years old: "So you're affected with FSHD and the constant not knowing as life progresses and this disease progresses. It's just torture."

**Trish**, who has lived with FSHD for nearly 30 years and is also a breast cancer survivor said she had "an amazing oncologist who calmly laid out a plan to beat cancer.... I later realized that having this plan was what got me through this nightmare. He was confident we could beat the cancer. And with his plan we did. FSHD in some ways is worse than cancer because there's no treatment, there's no plan. There's no guidance."

**Jack**, 60 years old, diagnosed with FSHD1 at age 32: "The progression of this disease has been difficult for me to accept. Three years ago, I was jogging, today I'm having trouble walking. The uncertainty about what's next is definitely affecting my quality of life."

## **FUTURE WORRIES: WATCHING DISEASE IN CHILDREN**

Heather: "Of my three children, two now have a diagnosis and I watched my third one like a hawk.... My fears for their future feel as though they'll crush me; both my girls have visible symptoms 20 years before I did."

## **FUTURE WORRIES: BECOMING A BURDEN TO FAMILY**

**Jeff B**: "It weighs on me all the time that I am becoming too much of a burden the more the FSHD progresses."

**Tim**: "I have burdens of, 'How am I going to have income for my family? As my job I used to do, I can no longer do.' I'm a burden to my wife, who has to carry this with me."

**Stuart** said FSHD goes back at least four generations in his family: "It was the toll that it took on my mom's health as a caregiver to my dad that I most noticed growing up. And that haunts me to this day and one of my biggest worries going forward is the effects of what this disease can have on the people who truly care for us. We want to be independent and be able to handle this disease ourselves. But the fact of the matter is all of our friends and family are very much intimately involved. And my constant fear is the effects that has on them."

## **COVID-19**

**Amy**: "The elephant in the room right now is COVID. And what I wanted to get across was that none of us have an exemption card, when you have one illness that you're not going to get another one or a second or a third. And with this disease, getting another illness and recovering from that illness



or having that illness exacerbate the FSHD back and forth makes any other illness that much more exacerbated. With COVID, one of my greatest fears is that if I come down with COVID even mildly.... An average individual might have been able to recover at home. If I had to self isolate, I cannot take care of myself on a full time basis, and that I would not be allowed to have these people responsibly come into my home, would mean that I would then be forced into either the hospital or nursing care system."

## TREATMENTS, OTHER MANAGEMENT APPROACHES USED, AND EFFECTIVENESS OF THESE MANAGEMENT APPROACHES

**Diane** described how she manages pain: "I try to do things to try to keep moving. I think that keeps the joints going. I find that when I do very low key working out, my back feels a little better. The ankles hurt no matter what."

**Stuart:** "Now all of my focus in life is doing everything I can with diet, exercise, and wellness, to hold out and keep the strength to be able to live independently and hold down or for a cure or something that can at least stop the progression. Because if they do stop I know I can live independently and not be a burden."

**India**, a 27-year-old woman diagnosed at age six, said that at about age 10 she started taking prednisone. "Although clinically this hasn't been proven to be effective, it reduced my number of falls and the severity of my fatigue. I was started on a low dose and would take it for about six to eight months then I'd be weaned off for a few months. Then I'd be put back on it with a slight dose increase. The prednisone suppressed my immune system putting me at risk for respiratory infections. I would get pneumonia sometimes twice a year... I'd also experienced rapid weight gain and trouble sleeping. I often felt highly energized with difficulty settling down or focusing. Once I was informed of the potential long-term side effects of prednisone coupled with many visits to the emergency room for pneumonia I decided the benefits weren't worth the risk anymore, so I've stopped taking the medication."

India also had a spine fusion to correct mild scoliosis and to prevent lordosis from creating permanent damage: "Having the surgery was not an easy decision. I was told that after surgery it was likely that I wouldn't be able to walk anymore because my FSHD had already progressed so much.... I chose to go ahead with it. The surgery successfully corrected my spine, but I lost the ability to walk independently. I spent about a month in inpatient rehab. I was able to get on my feet and use a platform walker, but mostly for exercise. Now, 12 years later I use my wheelchair full-time. I will say that without my power wheelchair my life wouldn't be what it is today. Although using a wheelchair also comes with its separate challenges, that doesn't change the fact that my body is constantly getting weaker making it difficult to use my arms. I tried Kinesio tape on my shoulders to give me better use of my arms. That didn't help much, it only left me with tape burns.



“FSHD has made it difficult to eat and breathe. I was given a BiPAP machine to use at night. I’ve had the machine for about five years now and I still struggle to use it daily. I sometimes wake up in pain with an extended belly full of swallowed air or I had to remove the mask because my nose and throat burned so much in pain from them being dry. I’ve had my machine settings changed several times. I’ve tried different sleeping positions, mask and humidifiers. In the days that I am successful at using it for more than five hours taking it off in the morning can be jarring. It feels as though that someone has suddenly turned down all the oxygen in the room. It takes time to adjust before I can move on with my day.

“To combat with my issues with eating I’ve changed my diet and the way that I eat. I avoid foods that are hard to chew or swallow and eat smaller meals to avoid fatigue, which can sometimes lead to aspirating and that list of foods is forever growing. While sometimes I use an adaptive spoon, pop up my arms or sit at an elevated table to be able to feed myself, many things that I’ve tried had been hit or miss and usually only work some of the time, like eye drops or sleeping goggles for the constant burning in my eyes. But things like hand splits or a soft back brace were more of a hindrance than a help. Oftentimes, the things that I try are difficult to adjust to making them less effective.”

**Jack G:** “I’m wearing a carbon fiber ankle foot orthosis brace (AFO) to help with the foot drop on my left foot.... It does help reduce the fatigue when I walk long distance and it definitely helps lessen my anxiety about tripping... I use a cane to help with balance.” I take a daily cocktail of supplements: selenium, zinc, magnesium oxide, CoQ10. I feel it helps me maintain the muscle strength, but there’s no real evidence of that and there’s no studies that are going to prove that it’s effective and they are expensive... For exercise, I’m riding a stationary bike 20 to 30 minutes, three or four times a week. I’ve also incorporated a light, but moderate exercise routine which includes using some weights, loop bands and then finishing up with some light stretching. I have experienced less fatigue and a positive effect on my quality of sleep, but I don’t feel that my muscle strength has changed any due to the exercise over the last one-and-a-half years.”

**Lexi,** age 26; diagnosed at age 12: “When I first went to a doctor many years ago, they told me that physical therapy was a good start. This is something that I did for many years from high school throughout college... Physical therapy helped with muscle weakness.”

One of the worst symptoms Lexi experienced was pain. Pain medications were ineffective so her doctors recommended steroid shots. “We felt that this treatment wasn’t successful and not worth continuing. Since then, I’ve had trouble finding something that actually helps with pain management. I’ve used THC pain cream, CBD cream, back braces/therapy and acupuncture.





Majority of those treatments really didn't do anything for me. THC cream is one of the only things that I've found that has helped with pain... it doesn't take away the pain, it just makes it a bit more bearable... The last treatment and the most successful treatment I've done is surgery. I had scapula thoracic fusion surgery... It's a risky surgery because you never know how your body will react. It could cause lung problems with the wires snap or pain from the wires digging into your skin, et cetera. But basically the surgeon fuses your scapula bones to your rib cage and uses wire to hold it in place while it fuses. The recovery was about eight weeks. And then several months of physical therapy. There are many pros and cons to the surgery, but for me, the pros outweigh the cons."

**Carden** said she also uses CBD: "I do take it orally. And that has really helped with inflammation. Since I also use a wheelchair, I've noticed that, because I don't walk anymore, that my legs and my ankles specifically get swollen. And when I do not take the CBD oil every day, I noticed that I get pain in my ankles and some swelling. So CBD has really helped with that. In addition, I do a lot of sound bath meditation therapies and stuff to really help to build that mental fortitude... the meditation and the sound baths really help with just managing the stress, the anxiety, and also just the depression that comes with FSHD."

**Jeff:** "I'm very consistent with daily exercise plan, consistency is slowly working to make me a bit stronger and more independent. I think to me, the biggest thing is to have a calm mind and be able to focus on being positive and continuing to move forward."

**Lance** said bicycling and yoga have helped him.

**Grace** said she takes supplements.

**Maureen:** "I've been doing almost daily Yoga for about 20 years or so and a meditation practice... I've been to PT enough over the years to know what they're going to tell me to do, and now I can do it in my Yoga practice. Keeping my mind focused and on the positives [through meditation] is essential to get through phase that are rougher than others as I see my disease progressing and there are more limitations physically, I have to keep myself mentally strong. So I think that's important."

**Kevin** said that about every three months he has "... ablation of the nerves in a different part of my spine to kill the nerves off, to help with the back pain. That does help quite a bit, but it just doesn't last long enough, insurance won't let them do it as frequently as I would like. So between that, I do have needling where they do put in some ketamine or something like that, also done to relieve the back spasms.... I also had the lower lid eye surgery where they lift the lid to where my eyelids can come down to, and that's really helped to relieve the strain on my eyes, especially when I go outside and it helps with sleeping because my eyes aren't open the whole time."



**Colette:** “There are many other treatments I have tried, ranging from custom 3D printed back braces to acupuncture. Nothing that actually treats or slows my worsening FSHD.”

**Dani:** “The best thing that worked treatment-wise is getting in the pool with my physical therapist.”

**Eduardo** has tried anabolic steroids, anti-inflammatories, cortisone, vitamins, and CBD. The only treatments that made a difference, were Ayurvedic treatment and yoga.

**George**, age 53, diagnosed in late 20s: “The thing that sort of keeps me going is exercise and it’s made a significant difference. I’ve been.... I was a bicycle racer for a long time until my mid thirties when my right leg and then left leg started to go, but I still ride regularly. I think it’s a great exercise, like swimming. It’s nonbearing, which I think is important.” He added that while he’s tried AFOs, eh find s them uncomfortable and fatiguing. Instead he uses high boots with a stiff ankle, which give him stability and allow him to walk better.

**Michael**, age 45 and diagnosed at age 23, said for many years he took diltiazem, a calcium channel blocker. “My folks ... did a lot of research back in the late 1990s after I was diagnosed that showed that blocking calcium in the muscles would prove to be beneficial. And there was since a study done... that showed that it was not effective. I did see some improvement... I certainly felt more fluidity in my gait, more confidence in my movements.... I started to have other side effects from the medication after a number of years, intestinal issues and lightheadedness, so I stopped.”

Other people mentioned taking creatine, selenium, zinc, and CoQ10 supplements, but did not know if they made a difference. Gerald wrote that nicotinamide riboside (Niagen) “has had a big impact over the last two years.”

**Anne:** “Massage therapy can be a great benefit for pain relief, increased circulation, improvement of attitude and reducing contractures.”

**Deborah:** “Cymbalta helps with pain as does massage. Golfing helps my two sides be better balanced. Restorative yoga helps maintain movement.”

**Robert:** “Swimming has helped me. I’m using power wheelchair full time, but I am still able to swim laps on my back. Swimming has given me great degree of independence. This is one of the very few activities I still able to do on my own. However, I noticed during the last couple of years, that it is taking me longer to swim the laps. That my strength and stamina are no longer there.”



**Jennie Lou:** “Acupuncture – this has changed my fatigue level tremendously – amazing! I can do more now than I have in years and have also been able to increase the reps of my PT exercises; belly dance – this is fun and has helped my core strength; meditation – this helps me feel focused and calm and positive.”

**Helen:** “Things I have done – moved to one level apartment/use of side arm crutches/buying a vehicle with better seating (and heat feature for seats)/re-arranging cupboard and closet storage. Purchasing a lift chair and laying back in it/taking rest breaks whenever I can.”

**Kathy:** “One positive treatment is reaching out and helping others whenever possible. Giving a friend a call, sending a text or email, paying kindness forward, being involved with meaningful events are small examples of living a meaningful life while coping with this disease.”

**Frank:** “I have been practicing kung fu and qigong to help fight back against FSH. When I first saw the neurologist at Johns Hopkins, after examining me, his comment was “whatever you are doing, don’t stop. These practices may not be doing a lot physically, but it is a mental battle that I am fighting, too.”

**Dana** wrote that hula hooping has helped stabilize her hip muscles.

**Mike:** “I’ve had excellent results with a specialized mesenchymal stem cell treatments. It’s expensive but make a massive difference with fatigue and energy.”

**Sandra:** “The only treatment that helps is the anti-depression drugs, which don’t help the symptoms but it helps with attitude and thoughts. Without it people really don’t want to be around me.”

**Amy:** “I used to be a golfer and I had to give that up. I’m an artist and I can no longer hold the brush to the canvas. People say, “Well, be inventive.” I come from a multiple generation family with FSHD, of cousins and uncles, and my father and grandparents. I have come to expect and learn how to be inventive and create adaptive equipment. There comes a point when none of that helps.”

## ADVERSE EFFECTS AND BURDEN OF MANAGEMENT STRATEGIES

**Carden:** “At 21 years old, I bought a scooter, a car with a lift, and ramp for the house that I lived in. The scooter collected dust though for six months because I knew people would treat me differently. Late spring of 2016, I took it for a spin and it felt almost like running again, though new challenges presented themselves due to the progression.”



**Carden:** “If I could describe my life in a single word, it would be adaptive. Every year poses a new challenge and I creatively find a work-around to get the job done, but despite the metal armor and badass attitude I wear on the outside I am scared underneath.”

**Chris S,** diagnosed at age 16, now 47 years old, also wears AFOs: “Although these AFOs helped me from tripping and falling I was very self conscious wearing them and didn’t want anyone to know I had them on. The AFOs also took some time to adapt to while driving and made walking up and down stairs more difficult. They helped me with some muscle groups, but also caused other muscles to gain more atrophy from less usage. ... At first, I did well, but when I turned 40 everything seemed to change. Even when I was wearing my AFOs, I began to fall. It seemed like everywhere I went there were obstacles. My doctors recommended physical and occupational therapy and after sessions in consultations with them realized we would need to renovate our house to make it handicap accessible. Those changes included an elevator lift, accessible bathroom, doors, cabinets and less steep stairs with special handrails and a ramp. Once the renovations were completed I felt more comfortable living in a safer house. However, as I saw throughout my life living with this disease there were trade offs. I now had an elevator lift, so I didn’t have to climb stairs and take the risk of possibly falling, but now my legs were getting weaker, so getting up from a sit to stand position was more difficult. I had more physical therapy to work on that sit to stand motion. Things would get better for a few months and then another muscle group would become weaker. I would use a cane for about year and then needed to move onto a mobility scooter and eventually a power wheelchair. Then, I needed to purchase a van with a ramp to accommodate the scooter or power wheelchair. I was happy because I could remain independently driving, however, as the months went on I then had difficulty moving my foot from gas to brake. I had a minor accident and after that situation I decided for the safety of my family and others on the road it was time to stop driving.”

**Kristin,** mother of a child with infantile FSHD diagnosed at age six: “The simplest outing for Kate has to be thought through versus just being able to go. Nonstop problem-solving, worrying and strategizing on all our parts just to leave the house... It takes a lot of time, research and chatting with many doctors and other patients to come up with game plan for your own situation. This has been a full-time job on top of the ones my husband I already hold... we strive to make sure Kate has as much of a normal childhood as possible, we have revamped and changed our lives drastically to do what we can to slow progression... We have revamped our entire way of eating with whole food and farm-to-table. It’s time consuming and expensive, but if it helps just keep her where she is today or slows progression we’re willing to endure. Our grocery bill has increased by over \$1,000 per month in order to get the right foods that have the right nutrition to help us build up Kate as much as we can.

“Kate has been going to physical therapy now two times a week for three years: The cost, however, \$450 a week per session, \$900 total. We also invested in a stationary bike that Kate rides four to



five times a week for half an hour.... We had to buy the high-end bike to fit her little body properly, which was a cost of \$2,500... We also have invested in Pilates and massage for Kate. Every other Wednesday night Kate goes for a two-hour session... Due to COVID, we had to stop. Dealing with the unknown year after year with what her life is going to look like, is mentally exhausting.”

**Ray**, father of two adult children with FSHD, described the struggles they had with deciding about surgery for their son: “We really wrestled with [whether he should have] spinal surgery to correct his lordosis, which was associated with his muscle weakness... we were concerned that he would lose his remaining ability to walk... we waited until my son started getting some ulcers... And once he started getting the ulcers, we knew we had to do the surgery. So it was a very difficult situation for us. He was 18 years old at the time and he knew one of the sequelae could be that he would not be able to walk again. So he had the spinal surgery and of course he was not able to walk anymore, so it was very difficult.”

There were some benefits from the surgery: “He was able to digest better. He did not have any of the ulcers anymore. He was able to sit up straight and didn’t have to constantly worry about his balance falling over while he was using the wheelchair... After the surgery, he did not require the personal assistance, so he was able to be able to live on his own as long as we had certain foods pre-prepared, which we did every week, while he was at college.”

**Maureen** had scapula fixation surgery: “It was a very difficult recovery... It was very long, it was very painful, much more so than I thought. I was doing a lot of pain medication, which I don’t love to take because I think as most people with FSHD know that we’re in pain all the time, so you have to moderate when you can take the medication.”

## DRAWBACKS OF CURRENT MANAGEMENT STRATEGIES

**Jeff J:** “The three areas would be effectiveness, side effects, and costs. Side effects would be liver damage. I take way more Advil than I like, but in order to deal with the pain throughout the day, I really don’t have a choice... Cost, it’s expensive. I mean, if you have to put a lift in your car or you have to buy a scooter, I mean, you’re talking five, 10,000 plus dollars, so this adds up very, very quickly. And then effectiveness, I mean, I’ve been riding a spin bike three days a week for 20 years. I haven’t gotten any better. Maybe I slowed down the progression of the disease, we don’t know, but I continue to do it for physical reasons and psychological reasons... “as you get older, it’s really hard to see the benefits of that. So you’re banging away and working as hard as you can, but at the same time you feel like you’re pushing a boulder up a hill. You’re just not getting.... You’re not making any progress. It’s a real test of one’s will to continue to put the time in not knowing if it’s really doing anything or not.”



**Colette** (35 years old): “At 12 years old, to treat scapular winging, my physical therapists taped my shoulders in place. The tape was embarrassing to wear at dance and swim practice and left a giant itchy rash. It did nothing to treat the disease from worsening. At 19, I got my first AFO to prevent falling and to help me walk. It was uncomfortable, hard plastic that required bulky athletic shoes, two sizes bigger than normal. Needing to wear an AFO felt like my autonomy over how I present myself in the world was being taken away, and it hurt so badly that my toes would become numb. Today, I wear a carbon fiber AFO that is more comfortable, but just as ugly. It does nothing to treat my FSHD from worsening.

“I also started wearing a corset-style back brace. Corsets were phased out of fashion for a reason. They are uncomfortable. Its interior, sharp metal bars slide out of place, bruising and stabbing my back and abdomen. The back brace does help me stand and walk for longer, but it does not treat my worsening FSHD.

“I had a lift installed in my car so I could bring my wheelchair. Because lifts are not considered a treatment, I had to pay out of pocket.”

She noted that she is fortunate to have the financial means to get what she needs, but “even with all of these things, this experience is still incredibly difficult to navigate and not good enough.”

**Janet:** “One of the hardest things for me is things like massage, supplement, exercise. We don’t know whether it truly works or not. Much of the evidence is anecdotal.”

**Dani:** “I have access to my neighborhood pool ... but there was no pool lift, so even if I wanted to use the pool to help me, I couldn’t get into it. It took me and my father three years to be able to get them to install a pool lift... We had to get the mayor of our town to email them and insist that they give me accessibility to something that I’m paying for [through the HOA in my community]. My father purchased me a floor lift. It’s an Indee floor lift. What happens is when I fall, he wheels it to where I am, and it slides under me and it lifts me into a standing position. Now, so many people do not have access to this because it’s so expensive.”

Several people mentioned that with gyms closed during the COVID-19 pandemic, they feel that their disease has progressed and they have been unable to do what is required to maintain function, mobility, and strength.

**Ashley:** “When facing severe pain daily, we’re sitting ducks for becoming drug addicts. Prescription pain meds are highly addictive. Don’t work for long, quit helping or devastate your liver and kidneys.”



**Susan:** “Our son began taking Oxycodone and Hydrocodone for his severe pain, which are helpful, but he worries about becoming an addict and tries not to take the pill every day. Only when the pain is just unbearable.”

**Amy:** “I have taken many brands of depression medications and the medications Lyrica, gabapentin, and tramadol for pain. All of the medications resulted in weight gains of 25 pounds or more. As my FSHD progresses I am even more unable to exercise to lose this added weight. The added weight makes it only harder to stand up, and to sit comfortably and is not healthy in regards to heart health and diabetes risk. I have only had bad experiences with physical therapy. I end up feeling more pain and also less stable on my feet than before therapy. So I have given up on physical therapy and currently available medications. The only thing that offers me comfort is a heating pad that I rotate around different muscle groups on my body. It only relieves the pain while in place, but it is something that I can use as often as I like without any side effects.”

## HOPES/EXPECTATIONS FOR TREATMENTS

**Julie:** “Any treatment that would stop the progression of the disease would benefit him so greatly. If we knew that the disease would no longer cause future muscle loss, that he could focus on building up the weak muscles that he does have.”

**India:** “I would like to see treatment that slows or stops the progression of FSHD so that something that works for me now will also work for me later.”

**Jack G:** “An ideal treatment for FSHD for me would be just slowing down or stopping the progression of the disease.” Jack described the difficulty of lifting his grandson for a hug. “Lifting up your grandchildren, taken for granted by most grandparents, has now become extremely important to me. If we can stop the progression of this disease, I’ve got two more grandchildren all waiting for Papa Jack to pick them up and give them big hugs and kisses. That alone would be a grand slam.”

**Chris S:** “If only there was some medication I could take that could help strengthen these muscles or at least stop the progression of this weakness so that I wouldn’t have the mental stress and anxiety of worrying about every step I would take with the fear of falling.”

**Kristin** (Kate’s mother): “A definition of treatment to us means something that either slows the progression or stops it. It would be a bonus and a dream to have her build muscles.... A huge piece of the puzzle is how you navigate life when you have a child with a rare progressive disease is the help of a treatment, no matter how minimal it’s affects. We don’t need something that solves the entire puzzle today, just something that keeps her who she is today would be life changing.”





**Lexi:** “Having a significant treatment for FSHD would really be life changing. I see two significant treatments that could help. I would like to see something that would stop progression of the disease. If I were to stop progression right now, I would still be able to walk in 10 years. I would still be able to smile, to get off the couch, to raise my arms, to hold my future baby and countless other things. I think about all the time how, if my disease keeps progressing at one point, I will no longer be able to hold my baby or play with them. Another ideal treatment would be regaining muscle. If there were a way that I regained certain muscles, that would drastically change my life. Sometimes I think about, if I could just have one more muscle in my abdomen, I will be able to do so much more and I would feel more confident. The abdomen weakness causes me so much pain and causes the way I walk to look different than others.”

**Jeff J:** “I would love to see therapeutics that would slow or stop the progression obviously. But more specifically for those of us who are still lucky enough to be able to exercise, being able to continue to do that for hopefully the rest of their lives has just huge, not only physical benefits, but it’s got a lot of also psychological benefits... When you’re able to exercise, you start to fire off these serotonin and dopamine reactions in your brain. And that really has a huge impact on attitude and behavior and mood and being able to lift your mood to take on the battles that we all have to take on every day. I think, it could pay huge dividends.”

**George:** “Then ultimately we’ll get to how do we regenerate muscle.”

**Colette:** “We need treatment that address the actual disease. Treatments need to stop patients from getting sicker and help us get better, and these treatments need to be affordable and accessible to everyone. Treatments need to help us regain strength, to not live in pain and to be able to enjoy life’s simple pleasures like picking up a child and cradling it in your arms, walking and scaling life’s mountains without having to worry about the disease burden.”

**Carden** added: “But I also want to add on to how the treatment will be delivered once we do get that and considering that. So is it oral? Is it an injection? Do you have to go to another location to get the treatment and what are those side effects after it? ... When I’m thinking about needles and injecting it, I have low dexterity in my hands. So I won’t be able to really inject anything into my muscles. So for me, I think it would be beneficial to have an oral tablet so that maybe it’s once a day, once a week, whatever it is, that would be the easiest way to take the treatment, if that could be possible.”

**Maureen:** “As a mother, there’s nothing I wouldn’t do to give my son a sense of future. And the uncertainty in this disease and the progression and not knowing where it’s going to end up or how you’re going to end up and what life choices you can make dependent upon that, are just so difficult.”



**William:** “For a normal person [to stand up], it takes about 200 muscles all kind of aligning together and helping you to get up. And for me, I’m probably at 20 muscles now. And so if I can just keep those 20 to still be able to get up and do some of that day-to-day activities, that would mean a difference between what future jobs I can do and what activities I can do with my friends and family.”

**Chareen:** “If I could take medication and I knew it would prevent me from being in a wheelchair. It would take a huge burden off my shoulders, knowing that my disease will not progress and I will not become more of a burden to my family. I struggle to walk. I struggle to pick up objects. I struggle to put plates in a dishwasher. I struggle to put my hair up in a ponytail. The biggest struggle is to keep a brave face in front of my family and my friends. And the next biggest struggle is the unknown. And I feel like having a medication to take away that unknown that I know that I’m not going to get worse would be life changing.”

**Janet:** “This disease is highly variable, and a drug that just makes slight improvements for one person might make huge improvements for another. A drug that doesn’t seem that helpful for someone who is very progressed in the disease might be helpful for someone barely showing symptoms. For this disease, even something that makes a slight difference is a worthwhile treatment.”

**Ellen:** “For [my husband] to be able to build muscle would be a huge game changer for him. To be able to easily feed himself, to sit up easily, use his arms well, and possibly even be able to walk again would be a true Miracle!”

## CLINICAL STUDIES

**Luke:** “I have also been desperate enough to take part in clinical studies. I had taken part in the ACE-083 study, which unfortunately did not have any positive outcomes or from what I can tell any negative outcomes, but the trial itself did end up failing and there were no improvements in strength. They were using that trial as an inhibitor to the [Myostatin inhibitor 04:15:05] that is naturally occurring within the human body.”

**Howard:** “Due to unknown rate of disease progression, it’s really difficult to tell if helping and/or to what extent. Lack of conclusive studies and therefore uncertain if helping to keep me stable strength wise, or if because it is considered a slowly progressing disease, whether my slow change in deterioration from year to year is simply where I’d be without any supplements, OTC drugs or diet changes.”



## APPENDIX II: Panelists' written testimony

### Panelist: CARDEN WYCKOFF

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Kurt, my mother said, have you noticed Carden's eyes don't close all the way shut when she sleeps? Kinda creepy right?

Eh, it's probably just her thing. I wouldn't worry about it, Kurt said.

My mother knew something felt different as early as four years old and so the journey began. Her eldest son slept with his eyes fully shut. So why not Carden?

"All right, class, you've got 60 seconds to complete as many sit-ups as possible. —Ready, set, whistle blows.—

Carden, come on! Sit up! Carden, what's wrong with you? Carden, 15 seconds left!

"I can't do it!" I screamed.

At the time, I was seven competing in a first-grade fitness test. I felt normal except I couldn't do a sit-up. I played midfield in soccer, first base in softball, center in basketball all through elementary school. I ran, walked, jump-roped just like all the other kids.

From when I was four through nine, the journey of being poked and prodded at doctors' offices began. My pediatrician knew something was different but he couldn't pinpoint it. We were referred to a local specialist who pinpointed the exact disease as soon as I walked in. Facioscapulohumeral muscular dystrophy. Facio what? FSHD. It's a progressive muscle-wasting disease which typically shows in adults, she said. To get a second confirmation, we recommend flying to Rochester to Dr Rabi Tawil to have Carden tested as well as the rest of your family. Results for only Carden came back conclusive. Spontaneous de novo mutation with no carriers.

Speaking to you now is this special snowflake.

I'm Carden Wyckoff, 27-year-old Atlanta native, wheelchair warrior, podcast host, techie, Appalachian Trail scaler, FSHD Society board member, world traveler, accessibility advocate and evangelist.

My journey with FSHD started in '97 and continues to be an epic one. I'll walk you through the lens I've looked through growing up to understand the progression of this debilitating disease. Though through it all, I continue to define all odds and break down barriers for people with disabilities.





Summer of 2007. Fourteen years old. My typical afternoon three-mile run felt off. I couldn't propel my legs forward like I used to. It felt as though they weren't mine. A mile in I collapsed, crying, on the concrete cul-de-sac. At the time, my favorite thing was stripped out from under me. I couldn't breathe. FSHD sank in. It clicked. Things would be different from now on. I peeled myself off the hot asphalt and walked home, tears dripping down my shirt. I told my mom it was sweat but don't think she believed me. I gave it another year-and-a-half, and 10th-grade flag football was the last time I ran.

My high school tenure turned into avoiding stairs and asking to leave class early because I had to use the "bathroom" since I couldn't walk across campus in 4.5 minutes before the next bell rang. I would hide class textbooks in the library because it was halfway between classes and they were heavy.

Stairs grew to be more of a challenge. First it was grabbing for a handrail; then it turned into using my right leg only to ascend stairs one step at a time. Falling down stairs in the house became a weekly thing, I eventually moved to the upstairs room.

My gait and lordosis became more pronounced. She looks pregnant. You look like you're dragging a dead Bambi. What's wrong with your hip? I ignored the mean comments, though they began happening everywhere I went.

I received handicap parking at 17 because walking distances was painful and exhausting. My brother began piggybacking me at music festivals; then sister shortly took on the challenge. Two ninja warrior courses and 82 miles on the Appalachian trail later....

I found myself modifying bending down, carrying groceries, getting dressed standing, doing hair, taking a shower, holding my cat to make up for the slow progressive muscle loss. I began holding my wrist with my other hand to lift my arm when eating. Chewing with my mouth closed became impossible due to the facial weakness.

My friends were talking about kissing boys and I'm worried about not tripping and falling on the graduation stage.

2012. Sophomore year of college. I fractured my skull after tripping in my parents' driveway due to the foot drop. Eleven days in the hospital and a tender spot on the back of my head the rest of my life. Shortly after, I received my first set of calf AFOs for foot drop. I could walk without tripping again! Three years later, four sets of AFOs after snapping them in half from tripping, and it was time for a scooter.

Winter of 2015. Twenty-one years old. I bought a scooter, a car with a lift and a ramp for the house I lived in. The scooter sat in my house for six months. I didn't need it. It was ugly. People would treat



me differently. Late spring 2016 I took it for a spin and it felt almost like running again. Though new challenges presented themselves due to the progression.

- 22 was the last time I used a regular stall bathroom because I couldn't stand without grab bars.
- 23 was the last time I drove because I couldn't pick my feet off the pedals.
- 24 I purchased a power wheelchair.
- 25 was the last time I stood to take a shower because I fell often.
- 26 was the last time I stood on my own....

If I could describe my life in a single word it would be adaptive. Every year poses a new challenge and I've learned to figure another way to do what I need to do to get the job done. But despite the metal armor and badass attitude I wear on the outside, I'm scared underneath. I think about the last time I will be able to chew solid food, as choking has occurred a few times. I worry about deteriorating into absolutely nothing but a bag of bones when all the muscle and fat dwindle away. I'm always cold due to my low weight, and winter season increases my immobility. I've given up writing with paper and pen because I have low dexterity. I wake up with my eyes dry due to lack of full closure. My smile and facial expressions weaken every year.

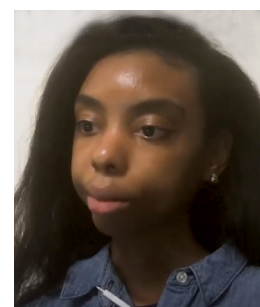
I believe we can beat this disease, and we need your help, understanding, empathy, and willingness to push drugs forward. I dream of the day I can walk again. The day I can dance, standing, at my wedding. The day I will be able to run again just like I roll now at six mph.

Thank you.

### **Panelist: INDIA PETERS**

Hello, my name is India Peters. I currently live in Arizona and I'm 27 years old.

FSHD is a progressive disease and my symptoms are constantly changing. It has left me no choice to be adaptable and come up with new ways to get through daily living. I've had to be creative and be open to new ways to cope with the symptoms and challenges that come with having this disease. It hasn't been easy, and I've tried my fair share of things from diet changes, hand splints, walkers, wheelchairs and other adaptive equipment.



In the beginning of my journey, I was prescribed prednisone. Although clinically this hasn't proven to be effective, it reduced the number of falls and the severity of my fatigue. It made my day-to-day easier and seemed to reduce some pain. But the prednisone suppressed my immune system,



putting me at risk for respiratory infections. I would get pneumonia, sometimes twice a year. I eventually had to make a choice, and I decided that the benefits weren't worth the risk. So I stopped taking the medication. Around the same time I had a spinal fusion to correct my mild scoliosis and prevent my lordosis from creating permanent damage. After the surgery, I lost the ability to walk independently. Now 12 years later, I use my wheelchair full time. I will say that without my power wheelchair, my life wouldn't be what it is today. But that doesn't change the fact that my body is constantly getting weaker, making it difficult to use my arms. Using a wheelchair also comes with its separate challenges.

Currently, in this part of my journey, FSHD has made it difficult to eat and breathe. I was giving a BiPAP machine to use at night. I've had the machine for about five years now and I still struggle to use it daily. And on the days that I am successful at using it for more than five hours at a time, taking it off in the morning can be jarring. It feels as though someone has turned down all the oxygen in the room. So it takes time to adjust before I can move on with the day.

To combat my issues with eating I've changed my diet and the way I eat. I avoid foods that are hard to chew or swallow and eat smaller meals to avoid fatigue, which can sometimes lead to aspirating. And that list of foods is forever growing. I also sometimes use an adapted spoon or prop my arms, or eat at an elevated table to be able to feed myself.

Many things that I've tried have been a hit or miss and usually only work some of the time. Like eye drops or sleeping goggles. The hand splints and soft back brace were more of a hindrance than a help. Oftentimes the things I try are difficult to adjust to, making them less effective.

All of these things are just short-term solutions to a bigger issue. And without treatment I will always be left to find new ways to deal with new or worsening symptoms. I would like to see a treatment that slows or stops the progression of FSHD.

### **Panelist: JACK GERBLICK**

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My name is Jack Gerblick.

I'm 60 years old and I've been living in Atlanta, Georgia, for the past 30 years. I grew up in northern Virginia, just outside of Washington, DC. I retired last year as a serial entrepreneur. I'm married and I have two daughters and three grandchildren. One of my daughters has tested positive for FSHD1, and my other daughter has tested negative. I'm the FSHD Society chapter director in Atlanta and also serve on the Board of the FSHD Society.





I was diagnosed with FSHD1 (mild to moderate severity) 28 years ago with a muscle biopsy procedure. The rate of progression of this disease for me over the past three years has been downright frightening.

I'm currently experiencing mobility and balance issues, weakness in my shoulders, and reduced upper-body strength. In addition to experiencing back pain due to lordosis and fatigue. These issues make it difficult for me to lift heavy objects, walk long distances, and raise my arms (particularly my left arm) over my head. I live in constant fear of tripping and falling due to the foot drop symptom on my left ankle/foot.

I'm treating my condition with the following:

Wearing a carbon fiber AFO brace to help with foot drop on my left foot/ankle. I'm a two-footed driver, so I can't drive wearing the AFO. Taking it on and off is doable but time consuming.

Supplementing the AFO by using a cane to help with balance.

Handicap plate so I can avoid walking long distances.

A daily "cocktail" of supplements. These supplements are expensive, and there is no real evidence that they are effective, but I still take them.

- Selenium 400 MCG
- Zinc 50 MG
- Magnesium Oxide 400 MG
- NAD+ (Nicotinamide Riboside) 100 MG
- Vitamin C 1000 MG
- Vitamin E 400 IU
- CoQ-10 200 MG
- Creatine HCl 750 MG

For exercise:

Riding a stationary bike for 30 minutes four times a week.

A physical therapy routine both in the pool (summertime only) along with using loop bands and relatively light weights. A 20-minute routine three to four times a week.

An ideal treatment for FSHD for me would be slowing down or stopping the progression of this disease. Slowing the progression of the disease even by as little as 25% would be like hitting a double in baseball. Stopping the progression altogether would be a home run.





My daughter and my now two-year-old grandson named Daniel, for the past year in our pre COVID-19 world, have been enjoying what we call “Fun-day/Monday” outings every Monday. We pick a location somewhere in Atlanta to meet. A nature center, Botanical Gardens, a playground, or a park; then all of us go out to enjoy lunch afterwards, chat, and get caught up. This has turned out to be the highlight of my week.

This particular Monday in late February 2020 we decided to meet at the local library. As I entered the library front door, Daniel recognized me and came running up with a big smile on his face and outstretched arms eager for a big hug from his Papa Jack. He was so happy to see me, as I was to see him.

As you can see from these pictures, lifting Daniel up was a struggle for me. Trying to keep my balance in order to keep him safe and not drop him on his head was my main goal. I was successful this particular day. Wait – now having said that – you’re probably thinking that I’ve dropped him on his head in past Fun-day/Monday outings. Just for the record, that has never happened.

If my condition progresses at its current rate, it will be impossible for me to lift up Daniel to give him a big hug. Lifting up your grandchildren, taken for granted by most grandparents, has now become extremely important to me.

If we can stop the progression of this disease, I’ve got two more grandchildren, Lucy and Caleb, waiting for Papa Jack to pick them up and give them big hugs and kisses. I want to be the best Papa Jack I can be for all of my grandchildren! That alone would be a Grand Slam!!!!

### **Panelist: JEFF BROOKS**

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FSHD is a Dream Crusher.

I started my life like all children, with dreams of being a sports star, firefighter, police officer, etc. As I grew I began to notice that I just didn’t develop like other kids. I was smaller, slower and more awkward compared to other children.

By the time I reached my late teens to early 20s, it was taking me a lot more time and effort to accomplish things, but I was a hard worker so things weren’t too bad. Except it took its toll on me. I’m in constant pain from all the wear and tear put on my body from having to compensate for the lack of muscle. Instead of living my golden years, it’s the broken years.

When I look back, I really enjoyed life from my early 20s through about 30. I thought this [FSHD] is manageable. Sure I won’t be no superstar but I am still able to go camping, to live sporting events, play softball, go bowling, go on vacations, etc. Things that made me happy.





I made a decision not to have children after watching all the troubles and heartache my grandfather and mother went through. Actually, it saddens me all the time that the difficulties my mom was going through I didn't understand, so I wasn't able to help her like I should have. I knew I had FSHD at that time, and there was very little known about it, let alone how to cure it. Actually, until the FSHD Society came along I had no idea where to get any kind of information or knowledge about the disease. Nowadays I feel not having children was a bad decision. I could use the help from them now, and I missed out on so very much. Everyone should be able to have the experience of having children and grandchildren, even though I am unable to lift and hug a child anymore.

When I hit my 40s, that's when FSHD started taking control of my life by taking away from me. The first couple of major things that I lost was the ability to climb ladders and stairs, which was a vital part of the work I did. If there had been some type of medication that would have stopped or even slowed the progression of the FSHD, I could still be working today. I went from earning \$70,000 a year to \$18,000. All the things that made me happy I was no longer able to do. For example, I went to a sporting event and had to use the restroom. I slipped and fell. Everyone just walked past while making remarks that they thought I was drunk. I haven't gone to a game since.

The depression and stress that is put on your mind is just horrible. I have lost all my confidence and stopped any kind of social interaction. I lived by myself for 10 years before I was introduced to the lady I live with now. I am worried that I will lose her. I am unable to make love to her because of the disability, and it weighs on me all the time that I'm becoming too much of a burden the more the FSHD progresses.

Over the last five years I have gone from being able to mow the lawn and go to the store by myself without any assistance to where the only yardwork I can do is from a sitting position on my scooter. It has become so difficult to do the simplest chores around the house, and when I go to the store or a doctor's office, if the wind is blowing, my body will freeze up because of the fear of falling and not being able to get back up to a standing position. I am at the point where if it is dark, snowing, or even raining, I won't leave the house. I have to pay someone to do any and all maintenance on everything. I no longer feel like a man. So when it comes to quality of life, I have to say I have very little.

With these troubling times, maybe the FDA could have a glimpse into my life, especially the quarantine part. This is my everyday reality, and it will be even worse in the next couple of years. I am unable to leave my house because if I fall, people can't just pick me up. So if those who are in quarantine will think about all the things they are unable to do now, and imagine that you will never be able to do those things again, that is what we in the FSHD family go through even without the coronavirus.

In my life I have been physically, mentally, and sexually abused. I was an alcoholic and a drug addict. I have lost everything I owned, but have been able to overcome these challenges and restore my life.



When it is all said and done, FSHD is what beat me. The slow progression of becoming a prisoner of my own body is becoming more than I can handle.

I know there are hundreds of thousands of people who have it worse than I do. All I am asking for is that the government of the greatest nation in the world and/or the FDA please help people with FSHD to have hope and the chance for a future.

Thank you for your time.

### **Panelist: JEFF JOHNSTON**

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My body feels like an iceberg floating south. As time goes by my muscles shrink, just like an iceberg melts as the weather warms. I've tried to fight it with every ounce of my body and spirit, but things continue to get heavier, the physical and mental pain is accelerating, and my optimistic nature is being tested like never before.



My name is Jeff Johnston, and I have FSH muscular dystrophy.

In my late 20s I started to notice changes in my body that didn't seem right. I was getting weaker, but I could still do all the things I loved to do, so sweeping my concerns under the rug gave me comfort.

It wasn't until I was 31 that I had to confront my harsh realities.

During an exam, my doctor referred me to a neurologist after seeing abnormalities in my upper body. Right then and there, hearing the doctor speak those words, I started to get nervous because I knew something was wrong with me. After a muscle biopsy, an EMG, and blood work, I was diagnosed with FSH muscular dystrophy.

As the disease progressed through my early to mid-30s, my legs and arms grew very weak. I wasn't able to pick my kids up out of their crib, or throw them in the air like many of my friends did with their kids. I had to sit down as my wife handed them to me, just like she did with their great-grandmother, who was fragile and weak. Playing golf – a game I loved – was no longer an option because my quads were not strong enough to support my weight.

Then came the falls.

They were unannounced, painful, scary, embarrassing. And as time went on, I struggled to get back up on my feet. I remember one time falling outside of Grand Central Station in New York City. It was during the morning commute. It was raining, and the wet concrete made getting traction extremely difficult.



My feet would constantly slip, and the more I tried to get up, the harder it got as my legs were burning with fatigue. Normally, kind-hearted New Yorkers would have stopped to help out, but since it was raining they just walked by, trying not to notice me. I looked like a cold, wounded animal covered in New York City dirt as I tried to do what so many people take for granted. I just wanted to stand up.

Fortunately, I didn't hurt myself, but I wasn't always so lucky.

Other falls have resulted in a pulled groin, bloody face, torn meniscus, bruised hip, bruised elbow, broken rib, broken finger, scuffed up knees, black eye, and intense headaches from my head hitting the ground.

Now, at age 50, walking is limited to a few steps inside my house. At this point it's easier to list the muscles that I still have versus the ones I've lost. I still have decent strength in my forearms and left calf. Otherwise, every other muscle is a shell of what it once was. This has made the simplest tasks difficult, if not impossible. Forget doing laundry; the clothes are too heavy. Forget making the bed; I'll end up on the ground. Forget loading the dishwasher; the plates are too heavy. And forget carrying a glass of water; I don't have the balance.

Growing up I was always an active and independent person. I liked to play sports, work up a sweat, go for a hike. Now, most of the things I'm passionate about have been taken from me. Now I get around in a scooter, which has been a major adjustment both physically and mentally. I used to have many good days where my energy level was high and my body felt loose and limber. Nowadays, pain is a constant companion. My lower back hurts from sitting so much, and the extreme hyperextension in my left leg results in knee pain after walking only a handful of steps. Being able to stand stationary relieves my back pain, but eventually my right leg goes numb and I need to sit down. Finding comfort is an ongoing battle.

You know, elderly people often fight efforts to have their driver's license taken away, or to move into a nursing home. And why is that? They fight these efforts because they don't want to lose their independence. Well, many people affected by FSHD don't have the luxury of being independent until their elder years. Some lose it their 30s or 20s, or even as early as their teenage years. This disease is wicked and cruel in many ways. But losing my independence is probably the most frightening and helpless feeling I have ever had.

Please help me get my independence back.

**Panelist: JULIE DYE**

Thank you for allowing me to speak about FSHD today. My name is Julie Dye, and my husband and I have four children and live in Corpus Christi, Texas.



In May 2016 I attended the end of the school year award program for my twin boys. They had just turned 10 years old and were completing the fourth grade. I was proud of their academic achievements, yet puzzled as to why one of my sons did not receive the award for physical exercise that all of the other students received. I looked into it and talked with the coaches, and learned that our son Noah was unsuccessful in doing sit-ups. My husband and I thought maybe the coaches didn't show him how or he just didn't try, and that we'd have to help him learn at home. To our surprise, Noah was incapable of doing a sit-up. It was not a lack of knowledge. We took him to his pediatrician and inquired about physical therapy to help strengthen his muscles. After conversing with his doctor, we realized that maybe there was something more going on and not just the inability to do a sit-up. Shortly thereafter we did a little research and realized that our son had had many symptoms of FSHD for years, and we just didn't realize they were all connected. He sleeps with his eyelids slightly open, he is unable to whistle or blow up a balloon, he can only drink from a straw if positioned in the corner of his mouth, he has a very small and flat smile, and he has winged scapula. Additionally, although he played sports ever since he was five years of age, his endurance was decreasing and he was not able to keep up with his peers. A neurologist genetically confirmed his diagnosis in September 2016.

Over the last four years Noah has attended physical therapy and was taught some exercises to do at home to maintain the muscles he does have. We learned early on that his strength would decrease throughout his life, and in an unpredictable manner. The physical therapist identified that he already has kyphosis, and his upper body strength and core strength were very poor.

After receiving his diagnosis of FSHD, we let Noah take the lead regarding his participation in sports. We didn't want to deprive him of childhood sports and playing with friends, but never wanted to push him to exhaust himself and his muscles more than he could handle. By the age of 11 he chose to not sign up for basketball or baseball due to inability to keep up with peers. He has since then joined Boy Scouts and the middle school band. Noah has enjoyed both activities, yet FSHD has continued to be a burden. He was unable to pass the swim test at Boy Scout camp, which deprived him of some activities, and he is unable to wear a multi-day framed backpack for an overnight camping trip, and unable to hike with gear for a five-mile hike like his fellow scouts.

As Noah is unable to purse his lips, he was unable to select the majority of instruments in band. However, he determined that he could play percussion, and he has become passionate about this



new hobby. As high school is starting in a few months for him, he has to face the reality that he won't be able to march in the marching band due to the inability to carry the drum over his shoulders.

There are many limitations that Noah has, and FSHD impacts his life in so many ways. As his mother, it is heartbreaking when classmates draw attention to the fact that he is unable to smile. Many photographers that we've used for family pictures have called him out, and have gone so far as to tease him and ask why he can't smile. Likely they assume everyone can smile, and little do they know it is not a choice for those with FSHD.

As I mentioned at the beginning, Noah is a twin, and he has two younger siblings. We are an active family and enjoy spending time outdoors. Over the last four years we have had to modify our activities and schedules to ensure that Noah is able to participate. Or we have had separate activities for our family to enjoy since Noah's poor endurance and strength impacts him daily. We recently went on a family hike, and four of our family members made it to the summit. Noah and I turned back down the mountain due to his exhaustion halfway up the mountain.

Lastly, a recent finding is that Noah is unable to raise his arms to comb his hair, unless he uses one arm to support the elbow of the other arm. At the age of 14 he is learning how to accommodate for his muscle weakness; however, I know his weakness and muscle loss will continue to progress.

As parents, like all parents, every decision made is done with the intention of giving our children a better future and giving them a leg up in life. Given the opportunity to find a cure for FSHD, we would do whatever it takes for our son. Although that is not likely in the very near future, a treatment is a possibility. Any treatment that would stop the progression of the disease would benefit him so greatly. If we knew that the disease would no longer cause future muscle loss, then he could focus more on building up the weak muscles and strengthening what he does have.

Every year we've noticed progression of the disease. Noah is on a path to real medical and physical limitations. If a therapy is shown to be effective, implementing it as soon as possible can mean all the difference between reversible and irreversible changes.

We thank you and all the people working so hard to find a therapy for FSHD, and we hope you can do all you can to give those affected a chance in fighting this debilitating disease.



## Panelist: KRISTIN ZWICKAU

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### TREATMENTS

My name is Kristin Zwickau, and FSHD became a part of our lives more than three years ago when our daughter, at age six, was diagnosed. She is what they refer to as spontaneous, as there is no genetic connection they have been able to make. She is also even more rare in the fact she has FSHD1 and FSHD2. The diagnosis took four doctors and nine months to finally get scientific results that gave us the answer to the battle we are fighting. Although her symptoms are severe physically, we cling to the fact that to date she has not shown any learning disabilities, Coats disease, hearing loss, or lack of lung capacity.



The day they told us she had FSHD is a day both my husband and I will never forget. It was at Boston Children's Hospital in the Muscle Clinic. We had never heard of the disease, so of course the words "there is no cure" just echoed in our heads. How could our daughter have such a severe disease that will rob her of mobility and there is no way to stop it, or just FREEZE HER? Questions swirled in our heads, that had and have no answers because there is varying degrees to how this disease affects everyone, and there is "NO CURE."

Somehow my husband and I had to figure out how to make her childhood and life the best it can be, allow her to understand her disease but not fear it, while we take all the fear on ourselves. It was time for us to lie down and give up – or FIGHT! We chose FIGHT.

Today Katelyn (recently changing her name to Kate) is a thriving nine-year-old despite the fact that she can't keep up physically with her peers. She can't run, walks slow, and struggles to go upstairs – all things her friends do without even thinking. Year by year as she grows, she is being robbed of things she loves:

**Skiing.** She was an amazing skier at the age of five, going down black diamonds and loving woods and jumps. Today she skies but only goes down the greens and has to hang on to my husband's pole for support.

**Walking.** She used to be able to walk 18,000 steps in one day, and now a big day is 13,000 and takes ALL day.

**Run.** She used to run and loved tennis. Now there is no running, and she can play tennis but the ball has to come to her; otherwise she will miss it.





**Bike.** She used to bike and lost her training wheels at age five; however, today she is on a tagalong attached to my husband's bike.

**Cooking.** She loves to do it but has to sit on the counter; otherwise, she can't get enough leverage to mix ingredients due to her arm and shoulder weakness.

**Carrying things.** She has to figure out how she can get her computer, books, lunch, etc., from one location to another – things her peers, again, don't even have to think about; they just do.

The list goes on and on, its heartbreaking to watch! The simplest outing for Kate has to be thought through versus just being able to go! Non-stop problem solving for her, and worry and strategizing on all our parts just to leave the house.

## DAILY LIVES/TREATMENTS

As I stated already, there is no cure/treatment or playbook for a child with FSHD. What they thought people should do 10 years ago is very different today. It takes a lot of time researching, and chatting with many doctors and other patients to come up with the game plan for your own situation. This has been a full-time job on top of the ones my husband and I already hold.

Over the course of the past three-plus years, however, we have learned feedback is key, and then we have to take that feedback and figure out if that works for Kate or not. As much as we strive to make sure Kate has as much of a "normal" childhood as possible, we have had to revamp and change our lives drastically to do what we can homeopathically and physically to slow progression.

**1. Nutrition:** It has been documented non-scientifically that high protein diets that are as natural as possible are best for patients with FSHD. Toxins in our food are extremely toxic for those with FSHD! We have revamped our entire way of eating with farm-to-table and whole food. It's time consuming and expensive, but if it helps to even just keep her where she is today or slow progression, we are willing to endure. Our grocery bill has increased by more than \$1,000 per month in order to get the right foods that have the right nutrients to help us build Kate up as much as we can naturally.

**2. Physical therapy:** Kate has been going to physical therapy for the past three years, and we know it works due to the baseline testing that they do. We have lost movement and strength in some areas while gaining in others.



## KATE'S WORKOUT SCHEDULE:

Monday-Thursday, 7:30-8:00, Kate rides a stationary bike. It's one of the few physical activities that is proven in FSHD to increase strength in the legs. In order to get a bike that was comfortable and fit her, we had to order a high-end spin bike. The cost is \$2,500 plus monthly subscriptions.

Tuesdays and Fridays, Kate has OT and PT with Spaulding Rehab in Boston. The cost is \$450 per session = \$900 per week.

Thursdays at school she meets with the school PT for a half-hour. In order to get this service, we had to put Kate on a 504 plan.

Every other Wednesday night Kate goes for a two-hour session for Pilates and cranial massage. The cost is \$220 per session.

We also have mats, balls, and weights at home to add and supplement Kate's exercise to keep her moving.

We live in a three-story townhouse. If there is not a therapy or a way to freeze her the way she is today, we will have to endure the cost of moving either to a high-rise with elevators or relocating altogether into a ranch-style house, meaning an uprooting of Kate out of her current school system where she has so many friends and an accepting, vibrant social life. It's a move we will only do if there is no cure solution in sight within the next two to five years.

As you can see from her schedule above, we have to incorporate a lot more activities into her everyday life than your average child. Most kids her age don't have to work out every morning in hopes of just maintaining function. They also don't have to add in three PT visits along with all her childhood activities.

## TREATMENT

Since the day we received Kate's diagnosis, we have been laser focused on a treatment. A definition of what a treatment means to us is something that either slows the progression or stops it! It would be a bonus and a dream to have her rebuild, but first and foremost is just slowing or stopping the toxin. Dealing with the unknown year after year of what her life is going to look like is mentally exhausting. My husband and I have no idea how to plan for her future, because we have no idea what condition she will be in five years from now if there is no therapy.

Do we plan that she will never have a job, and need 24-hour care and no college? Or do we plan that she will be able to be independent and have a successful career and a chance to live out her dream



of building a safe spaceship that can carry astronauts to far-off planets to explore? Our future and hers stay in limbo with so many unknowns – that if we had a therapy that at minimum slowed the progression sooner than later, we would be able to guide and plan for what her future looks like. Knowing is a huge piece of the puzzle in how you navigate life when you have a child with a rare progressive disease.

### Panelist: LEXI PAPPAS

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Hi everyone! My name is Lexi Pappas, I'm 26, and live in Massachusetts. I have been living with FSHD for about 14 years now, and it has definitely been a bit of a rollercoaster. Since I was first diagnosed, I was always looking for some kind of treatment or just something to help cope with the symptoms of FSHD.



When I first went to a doctor many years ago, he told me to start physical therapy. This is something that I did for many years, from high school through college. It is definitely not a real treatment, per se, for FSHD, but it helped to keep some of my strength during those years. The problem I often had, though, was that these therapists did not know too much about FSHD, so the exercises they had me do wouldn't always help. In college, I found a therapist who had taken the time to do some research on FSHD, and I could tell. Today, I still do a lot of the exercises she had me do. Physical therapy helped with muscle weakness, which is obviously the main symptom of this disease. I was able to gain a bit of muscle, but mostly it helped to keep some of the muscle that I already had. Something that came from PT and another type of "treatment" I did was going to the gym. This was pretty much exactly like PT, but I used a bit more weight and did exercises longer than I did at PT. Both of these are great at helping along the way, but they are definitely not a real treatment for FSHD.

One of the worst symptoms I've had from FSHD is pain. Starting in high school, I had very bad back pain. It stopped me from doing a lot of things in life, and I even had to quit one of the sports I loved because of it. Because of this pain, I wanted to find anything and everything to help with it. The first thing we tried was pain medication. The doctor prescribed medication that was stronger than Advil, but not too intense. These medications did not do anything for me, though. I didn't feel any different, and also preferred not to be on a pain drug to help cope. Because these medications did not work, my doctor recommended steroid injections for my back pain. The first time I did it, I did not feel pain for a few days or so, from what I can remember. I did it one more time and it had the same effect. We felt that this treatment was not successful and not worth continuing. Since then, I haven't tried anything that intense for pain. I have used THC pain cream, CBD cream, back braces, and massages. The majority of those treatments did not do anything for me. THC cream has been one of the only things I've used that has helped the pain, even if it's a little bit. I now have a lot of pain in



my neck, so the cream is something I use for that as well. Pain is hard to deal with, because most of the time it's never ending. Even when I go to bed, the pain does not stop.

The last and most successful treatment I've done is surgery. I had scapulothoracic fusion surgery in May 2015 and December 2015. One of the main and first symptoms of FSHD is scapular winging. My scapular bones have stuck out since I was about 12. It was always something that I was self-conscious of and sometimes caused me pain. I was also unable to lift my arms for more than a second or two without them falling down. My grandfather (who also has FSHD) researched this surgery for a long time and never wanted me to do it. But in 2015 he finally found a good surgeon and felt like the surgery was safe enough for me to do. It's a risky surgery, because you never know how your body will react. It could cause lung problems if the wires snap, or pain from the wires digging into your skin, etc.

Basically, the surgeon fuses your scapular bones to your rib cage and uses wire to hold them in place while they fuse. The recovery was about eight weeks and then several months of physical therapy. There are many pros and cons to this surgery. The pros, for me, outweigh all the cons. Some of the benefits are that my scapular bones no longer stick out, my shoulders do not slouch forward, there is little to no pain around my scapular bones, and some other small benefits. The negative side effects are that you do not have full range of motion and I have very large scars on my back. Overall, this surgery has been very successful for me as a treatment for my scapular winging.

Having a significant treatment for FSHD would be absolutely life changing. I see two ideal treatments for FSHD. In a treatment, I would like to see something that would stop progression of the disease. I think this would be life changing for so many people. I always hear from people that "if the disease would just stop progressing now, my life would be better." I think you can say this at almost any stage of the progression because it just always gets more severe as life goes on. Another ideal treatment would be regaining muscle. If there was a way to regain certain muscles, that would drastically change my life. Sometimes I think that if I could just have more muscle in my abdomen, my life would be amazing. The abdomen weakness causes me so much pain and causes the way I walk to look different from others. I often have strangers asking me if I'm pregnant or why I walk like this. So for me to gain muscle in at least one spot would be life changing.

**Panelist: MANUEL R. GÓMEZ, DRPH MS**

I was diagnosed with FSHD in college, but my case is extremely mild, and in any case, I am not here to talk about me. I am here to talk about my daughters, to give you a sense of what the impact of this disease has been on my two millennial daughters, and their mother and father.



My daughters are both, well, I would just say very good people, as well as accomplished and moving forward in their respective careers. They are not yet comfortable participating in an event like this, but I did get their permission to speak about them today.

Although my bad memory is a joke in my family, I distinctly remember the day I was watching my older daughter, Alina, then 15, in her softball practice, and wondered, with a stab of fear, why she had trouble running. Shortly after that, maybe even that same day or week, I saw both her legs and, although I still knew next to nothing about FSHD because it had barely affected me, it became very clear that her two thigh muscles were very different. Then it was from there to her primary doctor, then the neurologists, and her diagnosis, which was FSHD.

Alina is now 36, advancing in her career, living independently with her own car and condo. But mother and I have seen, in these two decades, the slow but relentless deterioration of her thigh, shoulder, and foot muscles. She already has severe difficulty walking. For any length of time or distance, she must use a wheelchair, and any conversation about a permanent wheelchair or scooter is extremely painful, both because of the fear it creates among us all, and because nobody knows what the progression of the disease might or might not be. She can barely go up steps, she has not been able to see the second floor of our house, she has great difficulty getting up from a chair, or even sitting down without falling. In her kitchen she can only – and barely – reach into the first level of shelves, and if she drops something, picking it up is out of the question if it is more than a “grabber” can handle. If she falls, she must crawl to a bed or other furniture to pull herself up. We especially worry about how long she will be able to continue to live independently in her condo or drive her current car, or when and how she will have to modify one or both substantially, changes which are expensive, and for which it is hard to obtain good advice.

Our younger daughter, Natalia, is 32, and until only a couple of years ago, we thought she had escaped it, although she had very mild symptoms, like me, and she had been diagnosed with the genetic test. She is also independent and recently completed a master’s degree. She had long been an avid jogger and urban cyclist, but she has now had to give up jogging and rarely uses her bicycle because of growing weakness in her legs. And she is very, very scared, because she has seen the decline in her sister’s condition over the years. So scared, in fact, that we fear she has put some distance between herself and her sister.



It is very hard to tell you all of this. The disease is relentless, it takes away my daughters' independence, and to a large extent darkens their future. It is an obstacle to their relationships with others, and I can't even imagine how they both adapt to the possible future loss of function, even their ability to walk or drive or live independently. I can't imagine how they adapt to the uncertainty of it all, and to the urgent expectation they must feel that the ongoing research will yield results that would halt the deterioration in their muscles – maybe even reverse it.

### **Panelist: CHRIS STENMON**

My name is Chris Stenmon. I am from Quincy, Massachusetts, and currently reside in Braintree, Massachusetts. I am 47 years old, and I was diagnosed with FSHD over 31 years ago at age 16. At that appointment, my neurologist said there was no cure and no treatments.



My parents were devastated. The term “muscular dystrophy” mostly inspired thoughts about “Jerry’s kids” and the Labor Day Telethon. One would think I would be as devastated as my parents, but I was actually relieved. At least my diagnosis gave me an explanation for why it was so difficult for me to make the throw from third base to first while playing Babe Ruth League baseball. The diagnosis justified the struggles I had building muscle while I was competing on the Boston College High School wrestling team. I finally understood why I had trouble smiling, which caused many people to ask me if I was “unhappy” or tell me I was “too serious.” As a teenager, I was very self-conscious of the way my body looked. I was not able to smile like other kids my age. I would not want to take my shirt off at the beach because I was fearful others would stare and make fun at how “strange” the scapular winging was on the back of my shoulders and arms. I would avoid going to the gym to lift weights with the guys because I knew I could not bench press as much as others shorter and younger than me. I recall those teenage years as my first experiences of seeing how this disease can affect not only my body, but also my overall self-esteem. If only there was some “medicine” I could take that could help me to smile like others or have my body look “normal.”

As the disease progressed and as I entered college, I knew I needed to find a profession that was not labor intensive on my body. I thought I wanted to be a psychologist. After receiving a D in psychology my first semester, I thought a business major might be a better alternative for me. Three-and-a-half years later I graduated with a business degree and passed the CPA exam the following year. During those college years and my mid- to late 20s, I began to realize that FSHD does not just affect the upper body muscles, but it also affects legs and ankles. I started to trip, lose my balance, and fall. It was like stubbing your toe, falling forward ... not being able to get your balance and having a “slow motion-like” fall. After several of these falls, I scheduled appointments with a physical therapist who referred me to an orthotist. These sessions resulted in me beginning



to wear ankle-foot orthotics (AFOs). This was my first experience with “durable medical equipment,” a concept I would become very familiar with in time. Although these AFOs helped me from tripping and falling, I was very self-conscious wearing them, and didn’t want anyone knowing I had them on. I would refuse to wear them in the summer when I was wearing shorts. These AFOs also took some time to adapt to while driving, and made walking up and down stairs a little more difficult. It helped with some muscle groups but forced other muscles to gain more atrophy from less usage. If only there was some “medicine” I could take that could help strengthen these muscles, or at least stop the progression of this weakness, so that I would look “normal” when walking and I would not have the mental stress and anxiety of worrying about every step I would take with the fear of falling.

My thought process at that time was that having or wearing something different would draw attention to me. I didn’t want co-workers or my clients asking me what was wrong with me. I was on the partner track at my firm and did not want anyone to treat me differently because of some physical limitations I had. Looking back on this, I see that I was putting myself at risk, and am I lucky something more serious like a head injury or broken leg did not occur.

As I entered my mid-30s, I was promoted to partner, got married, purchased a house, and experienced two of the happiest days of my life when my daughter Lauren was born and two years later my son Sean. When we first purchased our house, I was able to walk up and down stairs, walk without assistance of any durable medical equipment or devices, and drive the same as any “normal” person. When I turned 40, everything seemed to change.

Even when wearing my AFOs, I began to fall. I would slip on sand on the pavement, cracks on the sidewalk or street, inclines or declines. It seemed like everywhere I went, there were obstacles. My doctors recommended physical and occupational therapy. It was now at the point where we would need to renovate our house to make it handicapped accessible. We put plans together for a home addition to allow for a lift from the garage to the main floor, handicapped-accessible bathroom, bedroom, doors, cabinets, closet, and less-steep stairs with special handrails and a ramp. These costs amounted to over \$200,000. I remember being so frustrated because there were so few suppliers for certain items that the prices were much more expensive to make something “accessible” versus “for the general public.” This was yet another time in my life where I would say to myself, “Why can’t there be some medicine that can stop or at least slow down the progression of this disease?”

Once the renovations were completed, I felt more comfortable living in a “safer” house. However, as I saw throughout my life living with this disease, there were trade-offs. I now had a platform lift so that I didn’t have to climb stairs and take risks of possibly falling. But now my legs were getting weaker, so getting up from a seat in a sit-to-stand motion started to become more difficult. So then I had more physical therapy to work on the sit-to-stand motion. Things would get somewhat better





for a few months and then another muscle group would have pain. Then I could no longer bear weight on my right foot because of the sharp pain in my right foot and calf. I would use a cane for about a year, and then needed to move on to a mobility scooter, and eventually a power wheelchair.

I needed to purchase a van with a ramp to accommodate the scooter or wheelchair. I was happy because I could remain independent and could still transfer from the mobility device to the van driver seat. However, as the months went on, I then had difficulty moving my foot from gas to brake and had a minor accident where I rolled into another car while being stopped at a traffic light on a hill. After that situation, I decided for safety reasons I needed to explore hand controls for the van and other adaptive equipment. I researched different types of adaptive equipment, and the only product I could use due to the loss of strength that would continue to allow me to drive would cost approximately \$140,000. For this reason and for the safety of me, my children, and others driving on the road, I decided I would stop driving.

As you can see from “my voice,” FSHD has affected almost every muscle in my body. But its days are numbered. The “medicine” I have been hoping for all these years is coming to attack it. I have never been so optimistic as I am today for a treatment. I look forward to the day when FSHD patients do not have to suffer the struggles that were endured by previous generations of patients.

That is the reason I am here today. I have been knocked down physically, but I always get back up. I would like to end with a quote I recently read that I feel defines me and many other FSHD patients: “Toughness is in the soul and spirit, not in muscles.”

Or I could end with:

That is the reason I am here today. I have been knocked down physically, but I always get back up. I would like to end with a quote I recently read that I feel defines me and many other FSHD patients: “It’s hard to beat a person who never gives up.” I will never give up helping to find a treatment or cure for this devastating disease.



## APPENDIX III: Testimony submitted by email

### Anne H., Ontario, Canada

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I am the third generation within my family to have FSH muscular dystrophy. My grandfather had the disease as did my mother. My parents had three children and all three of us are severely affected by the disorder. The impact of FSH on an individual is profound. The physical deterioration often starts in the teenage years when self-esteem, identity and confidence are just beginning to form. Changes in facial muscles make you look just a little different, the shoulder and arm muscle deterioration make it hard to carry things, put your hand up in school, throw a ball. Often in your 20s you develop foot drop, making it impossible to run. Then muscles in the legs waste, causing numerous falls and injuries. Eventually you are unable to walk, stand from a seated position, roll over in bed, use a public washroom or take a shower. As muscle in the wrists and hands are affected keyboarding, holding a phone, writing, eating and dressing become laborious and eventually impossible. This is the scenario if you are fortunate as it is possible for the disease to strike early in which case you never walk, have difficulty hearing and speaking.

The physical changes are easily observed but the emotional and social impacts are more difficult to “see.” FSHD is an isolating “disease”. Because of stairs you are often unable to visit a friend or enter a family members home, walk hand-in-hand with your spouse, hug someone, hold a baby or run after a child. Even simple pleasures in life, like sharing a restaurant meal with friends, becomes difficult and embarrassing. Someone needs to cut your meat, a bowl of soup is impossible, food gets spilled down the front of your clothes as you struggle with utensils, and a coffee at the end of a meal through a straw!

These physical limitations are personally isolating. Friends and family must deal with emotional pain and grief. Those who aren’t able to watch this unrelenting decline pull away, further increasing the isolation.

The impact of FSHD is not just personal. It affects dozens of others.

Sadly for many a diagnosis of FSHD often leads to the heartbreaking decision to remain childless as couples feel they would be unable to physically or financially provide for a child or they fear passing on the defective gene.

The parents of a child with FSHD often face a lifetime of gnawing guilt over something they had no control over or perhaps even knowledge of. Even with genetic testing an accurate diagnosis is difficult to obtain. Patients are often misdiagnosed for years. Parents grieve each time they watch their child struggle with the simplest of physical milestones. They have a shadow of sadness overriding even happy occasions as they fear future losses.



A healthy spouse mourns the future they will not have but also the continual losses their partner experiences as another muscle wasted means the loss of yet another function. You watch someone you care about dying gradually before your eyes. You cope with grief daily as you witness decline and deterioration over decades. Everyone has tragedies in their lives. FSH patients are not immune to those but they experience additional layer upon layer of physical and emotional difficulties their entire life. The impact of the disease takes a devastating toll on the individual but the impact is far reaching and profound.

Over the last three months as the world grapples with the Coronavirus pandemic we have seen the consequences of social isolation. Entire communities have been devastated as they try to deal with a constant state of unpredictable loss. Around the globe research labs have been laser focused on a treatment and a prevention for Covid-19. Now imagine what we have experienced lasting the remainder of your lifetime without anyone looking for a cure or treatment. Until recently there were very few research labs focusing on FSHD. This adds the psychological burden of hopelessness to the “disease burden.” I do not draw parallels between COVID and FSHD lightly.

Living with FSHD is a lifetime sentence of struggle and isolation.

Please help us:

- acknowledge the seriousness of the disease
- recognize the large numbers of individuals with FSHD as well as those impacted
- obtain accurate physician diagnosis
- find a treatment to stop muscle deterioration
- develop a mechanism to correct gene deficiencies in utero or at birth

Without education, research and drug trials none of the above are possible. These are needed to provide hope in an otherwise hopeless situation.

Respectfully submitted July 31, 2020.

### Carolyn H.

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Pros and cons of treatments we have tried or are using now. What we would like to see in a treatment.

My name is Carolyn H. and I have FSHD, as does my brother Alex H. and my cousin Trish.

This disease has been in my family for generations and although we all have the same grandmother, we have very different symptoms have each tried various treatments.



My brother takes vitamins: C, calcium, D3, fish oil and has been doing physical therapy for over 22 years. He is in a wheelchair and worked an arrangement with the physical therapy school for students to come to his home and stretch him two times a week to maintain his range of motion and prevent muscle contractures. In exchange, the students get experience and learn first-hand about FSHD, which will better prepare them for their careers.

Alex has been taking albuterol and diltiazem, an anti hypertension and calcium blocker that increases blood flow to muscles for over 20 years – that is when he heard that these drugs could be beneficial to FSHD patients.

Each of these approaches have been beneficial for Alex's body, but none have stopped or impacted the progression of FSHD. He did feel less energetic when albuterol was not available for a period of time, but that was the only difference.

My cousin Trish also takes vitamins: multivitamin, CoQ10, B12, folic acid and has been seeing different physical therapists over the last 10 years. She is ambulatory so her physical therapy focus has been on her scapula and abdominal areas. Some PTs have done their homework on FSHD and those therapists help her re-attach her brain to her muscles and keep those connections live.

Trish also rode a bike in the gym until COVID closed the gyms, so now she rides her own stationary bike for 30 minutes every day.

As with Alex, Trish feels that the vitamins, PT, and stationary bike help keep her body healthy, but none of these have stopped or slowed the progression of FSHD.

I also take vitamins: B12, CoQ10, D, and fish oil when I remember and I have tried various physical therapists over the years. I am ambulatory, but have not had good luck with therapists and stopped going to them when they tell me that I will jump rope or be able to run again when they are finished working with me.

Because I was an avid swimmer and love the water, I swam an hour 2 or 3 times a week until COVID closed the pools. Since COVID, I try walking more and doing more floor stretching and exercises focusing on keeping my back stretched, tightening my core, and connecting my brain to my abdominal area. While these helped my overall health, the best treatment I have experienced was when I participated in the Acceleron Trial.

I was in the Phase 2 and then Phase 3 of the trial and received injections in my legs.

Not only did I see the physical change of the muscle showing where it did not before and most significantly in the 4 injection sites, but I also felt more stable walking than I had in years. For



example, I was able to walk on uneven surfaces, like my lawn, that I could not walk on for years without wobbling and falling. And getting up from a sitting position was easier as I suddenly felt those injected muscles engaged in a way they had not engaged before.

The physicians that gave me the injections also commented on how close to normal that muscle now was when they pushed against my foot as I raised it up from a toes pointed to the floor position and how they could visibly see the muscle in my leg when they could not before.

While this may not sound like a big deal and the Ace drug did not impact other parts of my body, it was a significant enough improvement for me to continue with the injections (even though I am very scared of needles) for the next few years had the trial not been terminated.

Alex, Trish, and I all agree – we want a treatment that will stop the progression of this horrible disease and are willing to risk a treatment that is safe and promising, even if it has not yet been proven effective. One example that comes to mind is Fulcrum's Losmapimod, but we know there must be others out there as well. We would not only accept that risk, but we would help assess the treatment by following all guidelines, keeping daily journals, providing regular results/reports, and anything else that may be requested for that specific treatment.

We are also reaching out to participate in clinical trials as they become available.

In summary, we need a treatment and we will partner with you in any way we can to get there as quickly as possible!

How the symptoms impact your life. What is the burden of living with FSHD?

FSHD is not something I am comfortable talking about; it is something that I wish would just go away.

One of my earliest memories is before kindergarten and my mom took me to the doctor to talk about FSHD. I remember hearing that there is no treatment and no cure, to which I added – no hope.

Because this disease has impacted so many people in my family, including my mom, grandmother, and brother, I never questioned if I had it; I only questioned when my symptoms would start.

I soon found out when we had this crazy Presidential Physical Education Testing in elementary school. I will never forget how horrible the timed 50-yard dash, pull-ups, and sit-up tests were because those were the ones I could not do, no matter how hard I tried and practiced for at home. Luckily, I don't remember the other tests because those were easier.



It was in high school when I noticed losing more – I could no longer jog, only walk. I could no longer lift things over my head, and I could not do some of the PE classes, like archery. I was grateful that I could still swim, play tennis, bicycle, play volleyball, so I focused on those and hid from the things I could no longer do. I also learned to appreciate everything I could do because I knew that one day I would not be able to do them anymore.

In college I got up the courage to see a muscular dystrophy doctor to check in both on my body and the latest research, but he only told me how weak parts of my body were and there was no treatment, no cure, and still – no hope.

It is now 30 years later, and this disease has become physically and mentally exhausting. Physically, I feel drained as if I just completed a major workout before I even start my day. Every morning I assess my energy level, and if I feel pretty good, I can plan a shopping trip or to get some things done before the afternoon when my energy fades. But if not, I will put those activities off for another day.

Mentally – and this is the part that I was completely unprepared for – my brain never stops working. Because I am no longer very stable, my brain is planning and assessing each and every movement and then constantly re-planning.

For example, when I walk – before each step I check the ground, is it level, are there any stones or gravel in the way, is it wet or slippery, where is the shortest path to where I want to go, are there any steps or curbs in the way, is there anything nearby that I can grab onto if I feel like I am going to fall, and if I do fall is it a safe place to fall? It does not stop there, my brain is always on the lookout for dangers along the way so I can avoid them: kids playing/walking can accidentally bump into me, dogs on a leash can trip me, wind can blow me over, people walking while talking on their cell can run into me, and any misstep or quick movement can put me on the ground in a microsecond.

I am exhausted, and I just need all of this to stop and go away.

I can't keep hearing that there is no treatment, no cure, and no hope. I need a treatment now, even if what becomes available first only stops the progression of this horrible disease. That would be an amazing start and I would take that the minute the drug is determined safe.

I am asking, actually begging, you to please make a treatment available today so I can keep the little I have left and keep my daughter safe.



## Christina E. and Leo S.

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My husband Leo and I wanted to briefly share our experience with early-onset FSHD. Our seven-year-old daughter Alexa was diagnosed a few years ago, but has had symptoms since birth.

She has Type 2 FSHD, and it affects just about everything except her spirit. She has significant cognitive, speech, fine motor, and gross motor delays. She has been hospitalized multiple times for pneumonia and respiratory illnesses, so much that she now has permanent damage to her lungs (bronchiectasis, or lung disease). She sleeps nightly and during daily naps with a Trilogy BiPAP ventilator. We have two 30-minute “meds” sessions each morning and evening, when she uses an airway clearance vest machine, Cough Assist, auction, nebulizer, takes two inhalers, and has daily and nightly doses of antibiotics. Eating has always been a major challenge, we think because her weaker muscles in her face and mouth just make it harder, so we end up feeding her about half of every meal to make sure she gets the calories she needs. She gets wonderful support and special education from her school, which describes her as having moderate to severe special needs, and gives her speech therapy, occupational therapy, physical therapy, adaptive PE, and specialized academic instruction.

Having FSHD has been hard for all of these reasons. But what’s even harder is knowing that this is a dystrophy and that, without treatments, the path may get harder.

For now though, we focus on the fact that Alexa couldn’t be a more awesome, resilient, wry, silly, and loving little person. We are hopeful that research and treatments will move fast enough to help our girl.

Thanks.

## Christy R.

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My daughter, who is currently 17 years old, was diagnosed with iFSHD at age three. However, she had symptoms from birth; we just didn’t know these symptoms were connected. She failed the newborn hearing screening and had her first sedated neuro hearing test at age six weeks to diagnose moderate sensorineural hearing loss. She began wearing hearing aids at six months of age. We thought it was a cute quirk that she slept with her eyes partly open. When she couldn’t blow out the candles on her birthday cake when she was two, we began to pursue a diagnosis.

Her upper body weakness, scapular winging, inability to smile and close her eyes completely were noticeable early on. She began using a wheelchair for school and when she would be required to walk for any distance, because she kept falling, at age eight. She had severe scoliosis with a spinal fusion at





age 12 that robbed her of her last bit of independent walking and standing. She has been wheelchair dependent since that time and requires full assistance with transfers. She requires assistance with daily activities such as dressing, showering, combing her hair, cutting food, and lifting anything heavier than about two pounds. She has difficulty writing due to weak hand and arm muscles. She has sleep apnea due to weak face and neck muscles, so when she lies down her upper airway is compromised. She uses a nighttime BiPAP machine. She often chokes when eating. She has severely weak core muscles and therefore has difficulty taking a deep breath, and is unable to effectively cough. She uses a cough assist machine to help her take a deep breath on days when she's feeling particularly tired or weak. Her physicians feel that as her muscle strength progresses she will require a feeding tube and mechanical ventilation. She has daily pain (which her doctors continue to deny is associated with the iFSHD) in all areas but mostly in her back and neck. She sees multiple specialists such as neuromuscular, pulmonology, cardiology, ophthalmology, audiology, ENT, and psychology.

Regardless of how affected she has been by this cruelly advancing disease, she graduated from high school a year early and is enrolled at our local community college. Unlike most college students, she is happy her classes will be online this fall so she can be comfortable at home and still learn. She wants to be a published author of a fantasy novel one day. She has many friends, and if she were able she would be involved in every activity she could find. However, pain and the need for a companion caregiver keep her from doing a lot of extracurricular activities. Simple things such as going to dinner with her fellow graduates isn't something she can do without someone who can assist her or who knows how to manage someone who is choking. She carefully chooses things based on her "is it worth it" scale.

Our small, four-member family has been impacted by this disease in negative as well as positive ways. Nothing we do looks like what a typical family does. We don't just jump in the car and go somewhere. It takes planning, effort, supplies, a wheelchair-accessible vehicle, and a service dog. We don't leave her alone for more than 60 minutes in case she needs something. But because of this, we are a very strong family unit, we have a large support network of extended family, friends and caregiver support; we are patient, and thankful for the small simple things in life and we take nothing for granted. Each day is a blessing.

Treatments to halt progression and or improve muscle strength would be life changing. We wholeheartedly support research and eagerly await promising new treatments. But mostly we look forward to treatments that will allow future children the opportunity to halt the progression before they are as impacted as my daughter has been.



## Cindy L.

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Thank you for the opportunity for me to share my story. It was the month between December, 1985 to January, 1986 – and the neurologist who diagnosed me was pretty blunt. Change your major so you can get a desk job, don't over-use the muscles, and think twice about having kids.

But – I was 20 – and when it came to my aspirations, I didn't listen. I'm a designer – and I worked in theatre costume shops off and on throughout my 20s – but it meant that I was on my feet for 10-13 hours a day, six days a week. Sometimes there were all-nighters – generally lots of coffee, not a lot of food. The one day a week I had off was often spent recuperating – sleeping a lot, and being kind of lethargic. The lifestyle was rough and I was getting weaker. I could feel it in my legs. I knew I had to pivot and find something else and I so I went to graduate school in my late 20s – I switched to graphic design.

I graduated with my MFA at 32. At this point I'm wearing AFO (those leg braces from the knee down) under my pants and walking slowly, but I figured, if I ignore it others will too. The interview and job offer I got from a studio in Chicago felt at first like a once in a lifetime opportunity. I was stoked – I really thought, they're looking past my weakness and focusing on my talent, you know? – but then I found out there was a catch. They didn't want to pay for my health insurance. I was supposed to work like a full-time employee, but I was going to be paid as a contractor – and could be let go at any time. So, I pivoted again. I had two other offers and took one with a local studio.

The job I accepted was on the second floor of a building without elevator access – and I was there for 13 years. A couple railings were the only modifications I needed, and my differences never got in the way of my job – until about 2010. I would wake up with a feeling of dread. I started getting anxious that I wouldn't be able to climb the stairs that day or fall trying. I'd literally prayed silently as I climbed the stairs. My arms were losing strength and I knew that getting out of my office chair was getting harder. I planned out going from room to room so that I didn't forget anything which would require retracing steps. The mental hoops I was experiencing every day from simply getting to and from work without fear was exhausting. If I'm being completely honest, leaving my position to start my own design business in 2011 was partially a move of necessity. One more time that the uncertainty of not knowing how long the strength I had would last compromised the next phase in my career.

So – I now run my own business out of my house with my husband's help (who is amazing by the way – got lucky with that one). In spite of my futile attempts with alternative treatments, my disease has continued to progress – I now use a wheelchair for long distances or when I'm feeling unsafe. I can still stand for an hour or two and walk short distances arm in arm with someone, but I can no longer get out of a chair on my own and require assistance in order to do so. Treatment for me would mean stopping the muscle wasting – giving me a little more time before the next fall,



the next weakened muscle, the next cyclical grieving process. A better treatment would allow me just enough strength to transfer from a wheelchair to a chair on my own. An amazing change to my life would be any additional strength and gaining back some of the independence I've lost. When deciding what level of treatment would be meaningful, please remember that my vote is for any safe treatment at all.

## Panelist: Dani L.

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### Panel 1 on the topic of living with FSHD: symptoms and daily impacts

My name is Danielle L. I am 31 years old. I was born and adopted from Santiago, Chile, and I currently live in North Carolina. I was diagnosed at the age of 12 by Dr. Robert Kurtzke, a neurologist in Fairfax, Virginia. I first noticed that I could not sufficiently raise my right arm above my head around the age of 11. Since I was an active child, my parents assumed I injured myself while playing rough outside. I always noticed in pictures that my top teeth didn't show as the rest of my friends', but I attributed that to having big lips. It became evident that I started to slow down, running, and my gait had changed. I began to get an extended belly, and I still couldn't raise my right arm over my head. Now when I say I was an active child, I played soccer, lacrosse, swimming, and diving, karate, bike riding, rollerblading; you name it, I tried it. So when we finally began seeing doctors, and there were a lot of doctors, I had no idea what to expect.

After my diagnosis and even more weakening of my muscles, I developed a slight limp, barely noticeable at times, but it became evident that I needed to make some modifications. I started gaining weight because my activity decreased so much, which was hard on my self-esteem. I had to deal with bullies in middle and high school. Not being able to participate in the same activities as your peers makes you stand out, and not in a positive way. I would lie about my disease, and I would say that I injured myself playing sports, and that is why I couldn't participate in physical education. In school, we didn't learn that there are people with disabilities who lead "normal" lives with jobs and families. I only knew what I saw from the media and learned from society's standards. No one told me that it was okay to be different, that I was not less, or a loser. I did have great friends that made sure I felt loved and included, we created fun memories, and I always felt like my disease didn't define me when I was with them, I was able to forget the fear of the unknown and just be a kid.

It is challenging to pick which symptoms are most impactful in my life because every day, there are challenges that require me to adapt or overcome. Some days I want to go upstairs, walk to the mailbox, get off the toilet without assistance, get up from a fall by myself, brush my dog, carry groceries from the car, roll over in my bed without struggling, the list is endless. There have been times I couldn't join friends and families on vacation or out in nature because walking was impossible



or the place wasn't accessible. I miss out on a lot of things with my friends because I cannot go upstairs to their homes, they do their best to meet me halfway, but the constant rejection from my own body takes a toll on my mental health. I would say the physical limitations can be less burdensome than the mental restrictions caused by this disease. Only in the past few years have I been able to find a therapist that I can talk to and explains and helps me deal with the grief of losing my abilities. She made my life more manageable when it comes to the toll FSHD takes on your mind.

When I usually wake up in the mornings, I have the most energy. I can get things done in the morning, like make my bed, clean my room, cook myself breakfast. I usually try to meal prep in the mornings due to my increase in energy so that I don't have to worry about cooking and cleaning later. Unfortunately, by 11 a.m., I already need to lie down, and sometimes that turns into a three-hour nap, and my day seems to have slipped away. Falling on the ground has a significant impact on my life. I am still able to walk around my house with a walker, and I want to continue for as long as I can. If I fall, my whole day and even the next day can be put on hold until the pain stops. I have tried to learn over the years how to fall correctly without hurting myself too badly, but I am not as agile as my cat. I am lucky now that my dad bought me a human floor lift, so when I fall, it slides under me and raises me to a standing position. Before that, if I fell, I would have to call EMS/FD to help pick me up, and usually, they would injure me more by picking me up wrong.

Sometimes I look back at the younger me and wish I could tell her that by the age of 30, things would be so different. That I would feel love and happiness even while using a wheelchair. A lot of people look at a wheelchair and think, "I would rather die than be in a wheelchair." That was me. Now I look at my chair with different eyes. I love it, I feel safe and can go so many places my legs can't take me. It has set me free. Even if there is no cure for me in my lifetime, I hope that with the advocacy work and bringing awareness to FSHD, I can help a young person with FSHD not feel so alone and scared.

### **Daniel Paul Perez, Past Chairman, President and CEO, FSHD Society**

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Respiratory issues such as sleep disordered breathing, insufficiency, and acute respiratory failure are of issue in FSHD patients. I've experienced both long-term chronic progressive issues as well as acute respiratory failure due to CO2 retention. I now use a ventilator.

Without doubt I am convinced that this is an unmet and misunderstood need in measuring disease progression in clinical settings and trials endpoints. Measuring tcCO2, etCo2, blood gas and metabolic measures, and correcting for insufficiency would be helpful. Improving pulmonary function is important to me and I would think one of the most accurate and sensitive measures for drug trials.



## Anonymous

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The effects of FSHD on my life

I am a spontaneous mutation and FSHD2 patient. I didn't learn this until adulthood, but in hindsight, it explains a lot of difficulties.

I was athletic until my early 30s. This does not mean that I was normal. I could do all the swimming strokes except the crawl because I couldn't raise my arms over my shoulders. I could also dive. I skied.

I lost my ability to jump in my early 20s. This sounds like a non-issue, but as the muscle loses reflex response, so goes the ability to jump, reach for an item on a shelf, and use your toes to stretch. As time passes, foot drop begins. First indication: scuffing the tops of shoes and tripping. At least there are AFOs and PLOs, which are plastic braces designed to alleviate foot drop, but the muscles continue to atrophy. Eventually, the braces and shoes become too heavy. The weight has not changed, but the strength to lift feet ebbs.

I was a textile designer and noticed that it was taking me more time to execute a project. I had to stop frequently and massage my hands. I lost my ability to hold a paintbrush and a pen. Can you imagine losing something as natural as the ability to write your name or jot down a note? As my fine motor skills went, my gross motor skills followed. I developed tennis elbow and golfer's elbow in the same arm, which decimated the joy of cooking. As the atrophy continued, all I can do now is direct a great meal.

FSHD affects every aspect of one's life, including showering and washing one's hair. I sit and bend over until my head is between my knees to finger comb my short hair.

Can you imagine getting dressed in the morning and contorting yourself to pull a top over your head or to put on pants? Buttoning a shirt is a non-starter, as your fine motor skills are gone. Zippers on coats are a 10-minute exercise.

You probably don't realize how many times you bend your elbow to lift a utensil to your mouth to eat. For me, it is a repetitive exercise during which I use up my energy and need a nap, so I fill doggy bags and finish later.

And then there is the getting up from a seat. Some days I need somebody to lift and stand me up.

Early in my gradual decline, I walked all over Manhattan. Then I needed a cane for balance. Then a walker, which slowed me down considerably. Now I use a power chair for anything over three blocks.

But I still use my ingenuity to adapt. For example, I do Pilates to keep me moving.



## Laura A.

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I'm lucky. On the spectrum of FSHD ability, I'd be way up at the top.

Problematic physical dysfunction did not manifest until I was over 50, and then for awhile, it was only obvious to me: no more high (or even low) heels; no more walking for exercise—which I'd been doing several times a week for decades—because it was too uncomfortable.

A decade and a half later, ambulation is awkward and unsteady, but still possible.

I see the muscle loss when I look in the mirror: skinny, sticklike calves and under-muscled thighs, regardless of how many minutes a week I spend pedaling the recumbent bike. Weakness has curtailed a lot of my activity.

What would meaningful progress look like to me?

No more episodes of muscle loss you can almost feel happening – extreme weakness and fatigue, muscle cramping and aching. Rebound from these episodes never feels 100%.

Progress in the development of bracing and orthotics – affordable, custom devices that are less obtrusive and easier to wear. One size doesn't fit all. Advancement in lightweight, durable materials. A way to try off-the-shelf devices so we don't wind up with expensive braces and orthotics we cannot wear or use.

Prescribing (and insurance coverage of) holistic approaches to therapy until pharmaceutical solutions are found – massage, yoga, aqua exercise, PT, whatever else brings relief and maintains physical function.

More affordable and accessible genetic testing and education for the medical community so it doesn't take years to get a diagnosis.

A cure. A drug or gene-therapy that would stop the muscle loss.

Treatment. Once the muscle loss is halted, therapies that return as much function as possible through compensatory muscular ability.

More and more hopeful research, until there is a cure.

I look at the people with whom I share this diagnosis and am in awe of how they cope. That includes my daughter whose defiance of FSHD's impact on her life is a model of bravery and resilience. Meaningful progress would be erasing her fears about the future. Stopping her muscle loss now so she can build from where she is and not wonder how much more muscle FSHD will steal from her.



We tell each other all the time how lucky we are. We have full and stable lives, otherwise good health, access to the care that exists for FSHD, the ability to participate in clinical trials, meaningful careers and community involvement, loving family and friends.

We are so grateful for the community of researchers, pharmaceutical developers, physicians, and support organizations that are working to find relief for us. Thank you!

## Noah E.

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I grew up playing sports, skimboarding, snowboarding, skiing, surfing, riding my bicycle, and skateboarding. Skateboarding was my primary mode of transportation. During my teenage years, my strength began becoming noticeably diminished. I came to a point where I could no longer run or play any sports. I was diagnosed in October of 2000. Because of my atrophied muscles, I have not ridden a bicycle or skateboard since 1998 nor can I walk up stairs without the use of a handrail, and has significantly reduced my ability to do manual labor.

I cannot get out of a chair without having the use of a high placed handle to assist. I am always falling down and have injured my knees, broken my foot three times, and have had numerous facial lacerations requiring sutures as a result. I have pronounced lordosis and scapular winging. My abdomen protrudes because of the weakening of my abdominal muscles. I am always in pain. I suffer from sleep apnea and have been prescribed a CPAP machine. I use a back brace, knee braces, and sometimes require using a cane or a walker to help my mobility. I am always tired. I do not take any medications. I have been involved with the FSHD Society and have been in two clinical research studies to advance research moving forward to a cure. I wish to have a treatment that will reduce *DUX4* in the near future. I hope you recognize the importance of your role in allowing these pharmaceutical companies to produce a viable option in medicine for the well being of the thousands of us who are unfortunate enough to be afflicted with the awful effects FSHD has on our lives.

## Ray J., Tucson, Arizona

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My name is Ray and I'm 61 years old. I live in Tucson, Arizona, with my wife, who also has FSHD. We are both power wheelchair users. Neither of us is able to stand or walk at all.

I was diagnosed with FSHD in 1993 at the age of 34. I have no family history of the disease, but the diagnosis explained why I was tripping and falling a lot and why I had lost all the athletic abilities that I had as a kid and early teenager.

I used a cane and leg braces for a few years. They helped a lot. I got my first (folding manual) wheelchair in 2000, age 41. I used it a little at first, but over about five years used it more and more.





One day in early 2005 I finally failed at dragging myself to my feet from sitting in the wheelchair. I have not stood, unless assisted by a machine, since then. Propelling myself in the manual wheelchair was difficult but possible. I adapted to life in a chair.

In 2007 I purchased my first power wheelchair. Suddenly I could keep up with the world again! But then also came the exclusion. With a manual chair, people would be happy to lift, drag, push to get you into places, up steps and the like. With a powerchair weighing 350+ pounds, that can't happen. So visiting family and friends in their homes is no longer possible. People stop inviting you to social gatherings, celebrations, and other events because you need certain facilities like ramps and accessible bathrooms. Your social group narrows to consist mainly of people like yourself, who need similar accommodations.

It becomes a form of social segregation. Or isolation in some cases. A viable treatment can fix that.

**I have given FSHD an updated name based on my own particular symptomology:**

- Facio
- Scapulo
- Humeral
- Abdominal
- Diaphragmal
- Tibial
- Femoral
- Radial
- Metacarpal
- Dystrophy

A mouthful for sure, but much more descriptive for my own case. I want to focus on the Metacarpal bit for a moment. I am a small business owner and part of my work, the part I love best, is electronic design, prototyping and engineering. This involves working with my hands on the workbench, assembling and soldering circuit boards and related electronic devices. This work requires some good hand dexterity.

I am now losing muscle mass and strength in my fingers. This is slowly robbing me of the ability to do one of my favourite things, which is also how I make income. Without income I do not know how I will be able to pay for wheelchair maintenance, home maintenance or the personal care I may need when I lose the ability to perform the functions for which you need your hands.



I am hoping for a treatment that would at least stop the progression of the muscle weakness. The sooner the better so that I can continue doing what I can do now; so that I can continue living independently with the adaptations that I currently have in place; so that I can continue making my own income and not have to rely on government support or charity; so that I can continue to live with dignity and purpose; so that I can continue my volunteer work advocating for people with disabilities.

Thanks again for the opportunity.

Kind regards.

### Christel R., South Africa

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#### **Symptoms of FSHD that have the greatest impact on your quality of life:**

The symptoms I have are all in the upper body. I cannot lift my arms above rib cage level. My symptoms presented themselves about 9 years ago, with severe winging of my right scapula. I have neck and shoulder pain constantly. In the last year, I have also developed lower back issues and experienced a lot of pain. At the moment, both my shoulder blades are winging badly and as a result, I am not able to lift my arms.

#### **Important activities that you cannot perform because of your FSHD:**

I cannot wash my hair normally. I have to place shampoo in one hand and bend scrub one side of head, then repeat for the other side. I cannot get dressed standing up. I have to lie on my back to put my shirts and dresses on and I put my pants and jeans on while sitting. I cannot bend down normally. While picking up a cup of coffee sitting on a coffee table, for example, I have to bend my legs in a squatting position and pick the cup up. I cannot lift anything heavy as there is no strength in my neck or upper back or arms. Getting in and out of the car is a struggle because it feels like I don't have the strength to support my upper body out the car, UNLESS my legs are in a position and at the height where they can support me. I cannot reach for things or lift plates or jugs from off the table, as my neck and shoulders don't support my arms anymore when they are carrying weight.

#### **Your greatest concerns about the impact of future progression of your FSHD:**

I'm concerned about my lower back muscles losing their strength too, as well as my core muscles. I am a positive and active person and have run many marathons and yet now I cannot run anymore, I have difficulty in walking longer than 15 minutes, so I am concerned as to how quickly this will deteriorate and how it will affect my lifestyle and especially my husband.

**Treatments you are currently using to try to manage your FSHD:**

Due to lack of finances, I have not had any treatments. I go to the gym to do weights for my legs so they can support me and I do core exercises at home.

What would a treatment need to do for you to consider it to have a meaningful impact?

To strengthen supporting muscles that are not atrophied, so that I can prevent other muscles from atrophying. I would also love to have treatment for my neck and shoulder pain.

**James C., Jenks, Oklahoma**

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Commissioners, my son and wife had FSHD. At age 23 my son had a difficult time walking, and weakness in his arms and legs. We lost him to suicide due to this destructive disease.

My wife suffered from FSHD and daily pain, and by age 50 required a scooter, and by 64 a motorized wheelchair. She also suffered from COPD. Diaphragm weakness caused by FSHD and the COPD led to an inability to expel carbon dioxide, which led to her death at age 69.

Commissioners, please do not allow families to suffer unnecessary deaths. Please expedite the approval of a drug which shows promise in halting the advancement of muscle weakness.

Thank you.

**William R. Lewis, MD, Davis, California**

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I have FSHD. I am also a physician and have spent my life caring for other people. This disease is slowly progressive and throughout my life I have had to adapt to the disease. I was diagnosed in my early teens and early on I was able to do most of the things I wanted to do. I was able to exercise, attend classes in college and medical school, and the disease was mostly a handicap and not disabling. As the disease progressed, I had to change my career plans from being a surgeon to being a cardiologist. I slowly had to give up exercising, because as the disease progresses, exercise changes from beneficial to harmful. Over time, I gave up doing procedures, then I gave up seeing patients (I was seeing the underserved), and I can now only sit at a computer screen and read diagnostic studies. The disease continues to progress and I find myself hoping that I can simply get up each day, get dressed, and get to work. Because of this disease, I will go from being a care provider to being a care recipient at an age way too young.

The disease does cause significant discomfort, probably because of the abnormal strains on our musculoskeletal system. I have had back surgery for herniated disc and continue to have chronic



back, neck and shoulder pain and muscle spasms. I also have thoracic outlet syndrome, probably due to the forward displacement of my shoulder girdle. This causes significant pain and numbness in my hands to where even typing on a computer is sometimes difficult. Because of wasting in my lower extremities, I now sit in a heavily cushioned seat designed for wheelchair patients in order to alleviate the pain and numbness caused by pudendal nerve compression. I use NSAIDs frequently for pain and am worried that they are causing my hypertension. To date, I do not have kidney failure, but that is also a concern. The muscle wasting of this disease also leads to insulin resistance and may cause early heart disease. Despite eating healthy and maintaining normal body weight, my cholesterol level and blood glucose are slowly rising.

As far as treatments are concerned, we all hope for a cure and a reversal of the muscle wasting so that we may all lead productive lives. If that is not possible, then treatments that arrest the disease or significantly slows disease progression would be our number two and three hopes. As a scientist, I want rigorous testing of potential treatments. I do not want therapies released simply to have something available, only to find they do not work, gave false hope, and actually slowed development of truly effective treatments. What endpoint we use to define effectiveness remains unknown. Reversal of the disease would be a miraculous endpoint and easy to detect. Slowing the disease will be a harder endpoint to detect and define. Ideally, treatment would be easy to administer, but we are all so desperate for effective treatments that we will even embrace treatments that are difficult or painful. The alternative is to slowly waste away.

Thank you for allowing me to give voice to this disease.

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### **Deborah W., North Carolina**

My son's swallowing is affected by FSHD and progressed to the point he requires tube feedings and nothing by mouth at the age of 33.

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### **Eduardo S., Brazil**

I am a lawyer in São Paulo, Brazil, and Vice President of ABRAFEU - BRAZILIAN ASSOCIATION OF FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY, or FSHD-BRAZIL, and I have FSHD....

I was diagnosed 27 years ago, in 1993, when I was just 20 years old.

More than half of my life I've lived and fought against FSHD.

Until the age of 20, i was a compulsive sportsman. I competed in tennis, squash, volley ball, full-contact (fight) and motocross.



I have always been sporty, always very competitive, very active and full of energy. I have always been extremely strong, physically speaking, which makes me very depressed with this disease, due to the fact that I am weak, unable to walk, dependent on other people, help....

I did all possible treatments, such as using anabolic steroids, cortisone, vitamins, medicines, teas, herbs, foods, cannabidiol....

I have also always done physiotherapy, gymnastics, weight training, hydrotherapy, massages, shamanic therapies, spiritual therapies, healing, self-healing, acupuncture, Chinese medicine, Japanese medicine, Indian medicine, Western medicine, yoga, Pilates....

The only one that actually had any positive effect was Ayurvedic medicine, in a treatment I did at a hospital in Coimbatore, Tamil-Nadu, South India.

I spent two months in hospital, with only vegan and organic food, natural medicines, made in the hospital, and massages.

It was three years in a row, each with two months of hospitalization, without being able to leave the room.

At all times, I came back to Brazil much stronger.

My body has been resting for three years, and I was due to return to India next December, but with the advent of the COVID-19 pandemic, the hospital is closed, and perhaps it may be I will go to India only in December 2021.

I have kept my daily exercises, with dumbbells, shin, rubber bands and balls, at home, because the gyms are still closed, because of the pandemic.

I weakened a lot this year, due to lack of the gym .... exercising at home is not as effective as at the gym.

Today, I walk with the help of two canes and, when I travel, I use a motorized wheelchair, which helps me a lot.

I'm afraid to use the wheelchair a lot, and get used to it and, thus, stop walking. So I walk daily.

I feel pain in the muscles, but totally tolerable. I exercise a lot, daily, which keeps me well. I use cannabidiol daily, without much effect, and vitamins such as Vitamin B, vitamin C, vitamin D, creatine, carnitine, arginine, taurine, leucine and a lot of turmeric, to lower inflammation.



I look forward to the results of the research, whether from Fulcrum, Bionano, Acceleron or any other.

I would love to be part of the clinical trial for any of these new drugs.

I'm sure I'll be able to stay on my feet until the FSHD TOTAL CURE is released.

In the meantime, I keep exercising, studying, helping others with ABRAFEU, following the news from FSHD Society and other associations around the world, always hoping to be healed one day, and return to normal life.

### **Robin S., Pennsylvania**

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I was diagnosed with FSHD when I was 14. Over the years I've functioned pretty well. I'm married with three grown children. My wife and I thought long and hard about whether to have children or not. In the end, we decided to go ahead. This was a good decision as I have three wonderful kids who've grown into great people.

I've lost a lot of function over the last few years. The FSHD, combined with blindness and injuries from a car crash in 1989 that forced retirement in 2015 at age 57. Life went on and I developed a music ministry, playing at area nursing homes three to four times per week until the coronavirus put all of that on hold.

My daughter had a baby last October, followed by the arrival of my second grandchild a month later when my daughter-in-law had a baby.

Being around the babies is great, but also very painful. If I'm in a chair with arms I can use to support my arms I can hold the babies. However, if they need to be changed, want to be walked or simply want to sit on the floor and play, I have to hand them off to someone else. I simply have the balance or strength to walk while holding a baby. If I get on the floor I can't get up without assistance. These are all things I did when my children were growing up. I looked forward to being a good grandpap for the next generation. Instead I feel like an observer.

A cure would be great. I'd also be thrilled if a treatment could be found to keep the condition from getting worse. My children don't show signs of FSHD, but I hold my breath every time they see a doctor. Of course, I hope that neither of my grandchildren ever have to deal with FSHD.



## Alejandra T.

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My name is Alejandra, and I am 35 years old. I was diagnosed with FSHD at around 13 years old. FSHD has a great impact in my daily life and defines everything I do physically and mentally. I am very insecure and have low self-esteem issues that I struggle to work on each day. I have no strength in biceps and upper back and cannot lift, hold or carry items by myself, and many times require someone else to do it for me. I can barely lift my arms above my head, and I am not able to reach for high things without assistance.

Even though I can still walk without assistance, my left leg began showing signs of weakness at around 30 years of age and I now have a foot drop. Any unevenness on the floor can cause me to trip. I have fallen many times, each being scary and unexpected. I cannot run or walk fast and must always scan the floor in order to avoid tripping and falling. I cannot run in case of an emergency. I cannot climb up or down the stairs fast, I must go slow and take my time, always minding my left leg who wants to drag. All this is being caused by the weakness and foot drop in my LEFT leg. How will I cope once my right leg becomes affected?

My greatest concern is progression and eventual loss of all abilities to care for myself. I don't ever like to think about the future or years to come and what's waiting for me ahead. I don't like to think about how time passing will result in more muscle loss, more weakness and more pain...slowly, randomly and unexpectedly. The ONLY thing that keeps me positive going forward, is thinking that with time also comes the possibility of progress in treatment. This fact alone makes me hopeful to think I might have a chance against this horrible muscle wasting condition. A future in which there will be some way to stop the progression, and stop it in time to gain back "some" control over my own body and muscles. Only this way I can think of my days ahead and not be clouded by FSHD.

I am 35 years old and FSHD is the reason why I chose not to have children. With debilitating muscles, I don't know if my body can go through a pregnancy and come out unharmed. My abdominal muscles are weak, how will it hold a nine-month belly? My arms cannot hold a newborn baby, much less an infant, so how will I care for a child? My legs aren't able to run; how will I run after a child to keep it safe? FSHD has deeply impacted my husband and me in this aspect of our lives.

I try to manage my FSHD with physical therapy and over-the-counter medication for pain. There is nothing that really works in preventing muscle loss. I wake up each morning thinking about my body and how debilitated it is becoming. The only way to cope is to ADAPT.

Adapting is key. I adapt and learn how to compensate for muscles that aren't working. Holding my arm up when brushing my teeth so I don't get tired. Resting my arms on a surface in order to comb my hair. Sitting down when putting on clothes so I don't lose balance. Wearing shoes with traction to





avoid slipping. Park close to where I am going to avoid long distance walks. Avoid stairs if possible. Avoid activities that require some sort of strength capable by a normal person. Adapting and compensating ... not really a viable treatment.

Stopping further muscle loss in FSHD would be major and very impactful. This would mean that I could be able to salvage whatever muscles are left undamaged and try to work on them. Stopping the progression of FSHD would be incredibly helpful and would most certainly have a huge positive impact in my life. It would mean that I could look forward with hope and not fear of my own body.

Thank you for giving me the opportunity and letting me participate and give my own perspective on how it is to live with this unforgiving condition. I am so very thankful for all the hard work and continuing progress being put forward by the FSHD SOCIETY and for fighting this horrible disease alongside us!

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### Rachel B.

I struggle on a daily basis with the challenges of having FSHD. This week has been a particularly difficult one where I have had numerous moments of losing hope as I live with less and less range of motion and strength and more discomfort and fatigue. I happened upon a recording of the forum on Facebook. I cried as I listened to many of the patients sharing their stories. This disease has slowly stolen so much from my life. The range of things that I am able to do lessens from year to year. As the disease progresses, it brings discomfort, pain, disfigurement, fatigue and loss of esteem. I would like to share my wish for a therapeutic to come to market as soon as possible that will stop further progression and that will help build muscle that has been lost. I would like therapeutics to be well researched which I know takes time, but to not be bogged down in paperwork etc. that can make this process take years. A therapeutic can not come soon enough.

I would be happy to share any additional information or to answer any questions to support this process and forum. Thank you so much for taking the time to address this disease.

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### Kate B.

Knowing that FSHD ran in my family, I was genetically tested prior to deciding to have a child of my own. The positive test result weighed heavily on our decision, understanding the 50% chance of having a child with the disease. Being unable to accurately predict the potential severity of the disease made the decision even more difficult. Additionally, anecdotal evidence showed that the stress of pregnancy could dramatically worsen my own symptoms, adding yet another factor to our decision. Ultimately, we chose to conceive naturally and have been blessed with a beautiful baby



girl. My worries now turn to the future health of our child, as I regularly look for any symptoms that might indicate a presence of the disease in her growing body. A cure or treatment of FSHD would mean that our daughter wouldn't need to struggle with the same decision as her parents when deciding whether or not to have children of her own.

## Mike G.

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My name is Mike. I was diagnosed with FSH muscular dystrophy when I was 55 years old I am now 66 years old. I have a right drop foot, and I been wearing a carbon fiber brace on that foot for 11 years. My walking has slowed down I have weakness in my hips and abdominal muscles. This disease has changed my life, and what I worried most about is if this disease will be passed on to my children. I consider myself lucky because this disease affected me late in life, but it is still a bitch. My Mother suffered with this disease, and so did an aunt and uncle. The sad part about this is they did not know they had it; they thought it was old age and arthritis. Hopefully one day a cure will be found!

## Fiona C.

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### **Symptoms of FSHD that have the greatest impact on your quality of life:**

FSHD has primarily affected my lower body, my mobility and balance is very bad, and I can't walk unaided. I struggle to sleep on a daily basis as a result of the constant pain I am in and get fatigued easily. The pain and limited mobility cause me daily challenges, everything takes longer, uses more energy, and I am unable to do the simple things like walk freely around my house or garden. I work full time still, determined that whilst my body is failing I can still be useful and contribute to society, but there is no doubt that this disease makes it difficult to do this.

### **Important activities that you cannot perform because of your FSHD:**

I used to enjoy lots of types of exercise, but can no longer participate in these. I used to love the freedom of being spontaneous and planning a family day out; now we have to plan everything in advance with meticulous research. Even going out with the family dog for a walk is no longer possible. I no longer go out for social events with friends. You lose your confidence quickly with FSHD, and feel guilty that friends and family have to adjust their plans so you can be included, so you can access a restaurant or venue or event easily, so it becomes easier to stay in. You get tired of the points and stares, people wondering why you walk funny, why you walk like a duck.

**Your greatest concerns about the impact of future progression of your FSHD:**

My greatest concern is what is round the corner for me, what will stop working next, and what if my son develops this. He is too young to be tested. currently.

**Treatments you are currently using to try to manage your FSHD:**

I have regular physio, use hydrotherapy and over-the-counter pain relief.

**What would a treatment need to do for you to consider it to have a meaningful impact?**

If it could slow down progression or stop it progressing further; that would a gift.

**Lauri P.**

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In the worst cases the disease is present all the time. Only a cure or an effective treatment could significantly change their lives and their mood. Loneliness is one major thing related to disability.

If there won't be a legal treatment, I am afraid there will be an illegal treatment. People are very desperate, as we heard. Any relief would be welcome. An illegal treatment is out of control, need of responsibility, need of ethics and moral, cover of insurances, taking care of side effects. For sure there would be a market for any kind of treatment.

Legislation must be up to date. Medical care and genetic technologies are developing rapidly. The progress will only accelerate in the future. Genetic and medical development must be within the reach of control.

Otherwise there would be fatal consequences.

Creating a cure or treatment does not apply FSHD patients only. It would be a giant leap for all neuromuscular disease research as well as all genetic research. That applies all humanity.

What I expect from a treatment: A drug that could stop or slow down disease progression. A drug that could help fight back against muscle waste and prevent muscle damage. A drug that could accelerate the action of patients' own stem cells to heal damage before it is too late. A drug that is safe, effective and has no negative side effects.

Some similar treatment like Spinraza or Zolegensma that are developed to treat SMA. Regenerating muscle fibers by using stem cells. Like organ transplant transfer muscle fibers from a donor though then a patient must take anti reject drugs for the rest of one's life.



So far there is no treatment. I am just doing physiotherapeutic exercises to try to maintain my current condition. It takes several hours a week and that is out of my free time.

Of course I am worried of myself. I was happier before I knew what causes my symptoms. Many times I wish I didn't know. I could live without the fear of future symptoms. A cure or a treatment would bring some value to FSHD diagnosis. So far it is just a dark report of one's future. I thought, what do I even do with such a diagnosis? There is no cure. But hopefully there will be in the future. It is strange. I feel both hopeful and hopeless about the future.

## Carolyn H.

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In loving memory of Joyce Hakansson (March 1940 – November 2016)

Despite the many challenges and hardships brought on by FSHD, my mother, Joyce Hakansson, was a gifted, innovative entrepreneur, visionary, strong leader, and used her many creative talents to make this world a better place. My mom had an energy and presence about her; you knew when she entered a room as people naturally gravitated to her loving and nurturing personality. And she was my strong role model as she battled FSHD which unfortunately cut her achievements and eventually her life before her time.

My mom was gifted - she graduated from High School at 16 and graduated from UCLA at 20 with a BA in history, a minor in physical education, and a Secondary Teaching Credential.

In the late 1970s, my mom was the director of the Computer Education Project at the University of California, Berkeley, Lawrence Hall of Science and convinced (a barefoot) Steve Jobs to donate 10 Apple IIs, which were then taken around to schools in an Apple Van for kids to try their hands on. In 1979 she was hired by Children's Television Workshop to develop the Computer Gallery at the soon-to-be-opened Sesame Place in Langhorne, PA. Despite FSHD starting to take a toll on her mobility, she still commuted across the country to work with both the artistic folks in New York and the engineers and Steve Jobs in the Bay Area.

After Sesame Place opened, my mom returned to California and started using a scooter. She found a way to independently get her scooter in and out of her 80's Jeep Cherokee and started a company in Berkeley, CA. Joyce Hakansson Associates, Inc. (which ended up changing its name to Berkeley Learning Technologies, Inc. and finally to Theatrix Interactive, Inc.), to develop fun educational software for children. The company also developed products for companies like Texas Instruments, Sony Japan, and Hasbro. Several programs were translated into foreign languages and some were used in the Swedish pre-schools. She even gave a copy of her software to the King and Queen of Sweden for their three children.



Her company also produced the interactive exhibit at the Hubert Humphrey Museum at the University of Minnesota and a music program with the Juilliard School. My mom co-founded the not-for-profit Learning Express with Bruce Murray, who was head of the Jet Propulsion Laboratory in the early days of the space program (and co-founder of the Planetary Society), and John Gardner, former HEW Secretary in the Johnson administration and founder of Common Cause. One year my mom's company, Theatrix, even won three Codies (same concept as an Oscar) while Microsoft won one!

Despite the challenges of traveling with a scooter in the 1980s, my mom did not let that hold her back. She was invited to speak at universities around the country, from Harvard to Stanford and Berkeley, was a keynote speaker in Australia, and served on a panel in Cannes, France. She was also active in community affairs, including service as executive director of the Alliance for Technology Access. She testified on intellectual property before Congress, gave talks at the Department of Education, and even met President Clinton in the White House.

By the time my mom was 46 she was forced to rely on her scooter and shortly thereafter, an electric wheelchair, and had to cut back on her traveling and activities. Then at the young age of 59, FSHD put her in the hospital for 40 days and she required a full-time live-in caregiver thereafter. Despite these physical challenges, she continued as CEO and creative director of Theatrix for another 10 years retaining the innovative spirit and positive and engaging outlook on life's opportunities for which she was known. And she remained as creatively active as possible, including co-chairing FSH Society fund-raising events until FSHD ended her life at the young age of 76.

My mother was the strongest, most energetic, positive, creative, and out-of-the-box thinker I have ever known and did everything in her power to not let FSHD get in her way or bring her down, but I saw this disease progressively and dramatically shrink her world, completely suck the life out of her, and fill her with both guilt and grief as she saw the ugly progression of FSHD in me and my brother. It was FSHD and the obstacles this disease creates that kept my mom from reaching her potential, her dreams, and broke her body and eventually her incredibly strong spirit way before her time.

**Here are a few of her health challenges brought on by FSHD that I remember:**

**Mental anxiety/depression** – As your body progressively gets weaker, you can no longer trust it to keep you from falling, be able to walk up the few stairs or a curb to get into a mall or store/restaurant, find an open accessible bathroom when you need it, or find a place to sit and rest while you recover your energy from walking a few yards. And when you are away from home using a wheelchair, you need accessible parking (which people without disabilities take), accessible access, and an accessible bathroom. The constant planning and re-planning required to make sure your body does not fail you or you have the access you need is exhausting and the anxiety that goes with it sucks the life out of you. You find it easier and less stressful to avoid it all and just stay home. The



more you stay home, the more isolated you become and depression sets in. As my mom's FSHD progressed to where she could no longer drive or use a bathroom without being lifted out of her wheelchair, she stayed home which was very hard on her extroverted personality.

**Broken leg** – My mom fell one day after loading her scooter and broke her leg in several places which required surgery, pins, and a full leg cast along with being confined to bed. That was the last time she walked; after her leg healed she was confined to a wheelchair due to weakness from the break and FSHD progression from being bedridden.

**Fused vertebrae in her neck** – Due to weakness in her core from FSHD, my mom often used her neck to help lift her body. This resulted in damaged neck vertebrae which in turn would have caused paralysis without the fusion surgery. After surgery she was sent to a convalescent hospital for weeks for physical and occupational therapy.

**Bedridden due to broken wheelchair** – Most people don't know the challenges wheelchair users experience with their equipment. Besides being insanely expensive with very little insurance help, wheelchairs break and there are very few people and companies that repair them. I recall several times when my mom's electric wheelchair broke and it took several weeks to months for the wheelchair people to have time to fix it or to get a replacement part. Wheelchairs are customized to fit the user's strengths and weaknesses and due to her FSHD, my mom required a very specific setup. While her wheelchair was broken and waiting for repair, she had to spend her days and nights in bed. I remember a few times when my mom's home-care assistant put my mom in her electric wheelchair and pushed her – literally several hundred pounds of weight of wheelchair alone – out of her bedroom so my mom could get out of bed and her room for a short while.

**Core weakness and pulmonary issues** – Because of her weak core due to FSHD, my mom wore a corset to keep her back straight and stable for as long as I can remember – I think she was in her 30s when she first got it. The corset was tight so she could stay straight, but it also impacted her lungs and made it hard for her to cough and clear her lungs. She was at high risk for choking and pneumonia, especially during allergy and cold/flu season. More often than not, she ended up in the hospital if she got a cold so they could help clear her lungs for her and prevent pneumonia.

**Tracheostomy** – My mom had a bad cold and was having difficulty coughing to clear her lungs. She went to the hospital and their pulmonologist told us that due to her blood pressure dropping so low when she coughed and her weakness from FSHD, she would die without a tracheostomy, which they performed against her wishes. What should have been a short-term solution, quickly became permanent. She was put on a ventilator and then sent to a horrible rehab center for 6 weeks to try to wean her from the ventilator. The rehab center gave her no support and because my mom could not move without assistance, she was completely neglected while she was there. The whole



family fought to get her released, but it was not possible until her 6 weeks of Medicare payments ran out – and only then was she allowed to return home with the ventilator and tracheostomy she never wanted. I spent countless hours from my home in Oregon with the insurance and medical supply companies working through issues and weekly deliveries of oxygen tanks, tubing, cleaning materials, and other parts that were enough to fill a large closet. I had to create an excel spreadsheet to track the part numbers of her supplies along with when they were used and delivered to make sure she did not run out of anything. Finally, after several years of hard work, my mom was able to convince a pulmonologist to remove the tracheostomy. My mom told me later that the tracheostomy was never needed and if we only had someone on our side that could explain FSHD, this would have never happened.

**High expenses** – Having FSHD is incredibly expensive! Because my mom's medical expenses were so high with her basic medical needs and hospitalizations, my dad found special medical insurance to help cover the expenses. Despite the special insurance they still had unimaginably high out-of-pocket expenses for things like wheelchairs, wheelchair adaptive vans, wheelchair/van repairs, hospitalizations, medications, medical supplies, caretakers (often needed 24/7), house modifications (lifts, etc.), and more. Financial tradeoffs had to be made including not doing home repairs and my father driving a 20+ year old car.

**Death** – My mom was an amazingly sharp and brilliant 76-year-old with more drive and experience and dreams to do great things than most people will ever have, but FSHD had taken over her body leaving her unable to move or adjust, unable to sit without being strapped into her wheelchair, barely able to move her thumb to control her wheelchair joystick, unable to feed herself, unable to get comfortable in bed, unable to brush her hair or teeth, unable to wipe her tears when she cried, unable to scratch an itch, and completely 100% dependent on her care-givers for each and every aspect of her life. No matter how hard my mom fought, FSHD stole her independence, turned her into a quadriplegic, and when she got that last lung infection, FSHD had destroyed so much of her body that there was nothing left for her to fight the infection with. My mom fought this disease with everything she had every day of her life, but in the end, FSHD won and her body failed her.

I would like to share what my mom wrote in the FSHD Songs In The Key of Steven Blier program in 2014 as I feel it is also applies to the work the FDA is doing for FSHD:

“Thank you all for making this evening possible. Words cannot express our appreciation and gratitude for your generous donation of time, talent, hard work, and assets to meet the goal of eliminating this despicable disease. I can trace my link to FSHD through five generations, and it probably went back further than that. Now, for the first time we are seeing real progress in identifying the cause of the disease and with the help, and generosity of people like you, a treatment





and cure are a possibility. Then, future generations will not suffer the limitations and frustrations that so many have and are experiencing.”

### Kristen L.

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I, Kristen L., am writing to you as the mother of a multi-disabled young boy named Noah.

Noah is only seven years old and has infantile onset of facioscapulohumeral dystrophy, or FSHD. He is severely affected with this disease – the result of a sporadic mutation that occurred very early in my pregnancy. Often, people consider this disease only as a hereditary one, but Noah is the first in both paternal and maternal families.

Infantile onset usually indicates severe weakness, early wheelchair use, and in some instances, hearing and vision loss, which is true for Noah. Noah is also non-verbal and cognitively much younger in age. I personally believe this is related to being severely affected by FSHD.

Noah was born with a bilateral sensorineural hearing loss, leaving him with only some hearing in each ear. This was the first diagnosis that would eventually lead to FSHD. Later, when Noah was only nine months old, we learned that the blood vessels behind his retinas were leaking fluid, detaching them. This condition is commonly referred to as Coats disease.

We were told by an ophthalmologist that Noah was blind in his right eye. For the past seven years, Noah has had to undergo laser eye treatments and surgeries to both eyes. Sometimes, the treatments occurred every few weeks. Though the doctor was hopeful in saving the left eye, unfortunately, Noah’s vision could not be saved. He is totally blind in both eyes and can only see light if he is in a dark room with a flashlight shining directly in front of his eyes. All future treatments and surgeries are to maintain the health of his eyes. Noah has, and will continue to use, glaucoma drops for the rest of his life. If the pressure cannot be controlled, it is possible that one or both eyes will be removed and replaced with a prosthetic.

The number of doctor appointments related to FSHD, as well as occupational therapy, physical therapy, and speech therapy, made it impossible for me to continue working. In addition, Noah frequently gets sick throughout the year and needs to remain at home, often for a week at a time.

Having a blind child with both a hearing loss and muscular dystrophy presents a huge challenge in teaching and entertaining him. Overtime, I have come to accept that he will never be independent, have friendships, or regain vision. Maybe he will never talk. However, as a mother, I cannot accept him living a life without joy and will do everything in my power to help him find it each day.



There are few things that really make Noah happy, and one of those is being able to jump on his small trampoline while holding onto the attached handlebar. It may seem simple, but for him, it is something he can do independently and whenever he wants. I do not know how much longer he will be able to do this activity; even thinking about him not being able to jump breaks my heart.

Time is of the essence in providing a treatment or drug that can slow or completely stop this awful disease from progressing further. I am desperate to keep Noah jumping and as healthy as possible. I am pleading with the FDA to please consider approving the treatments for FSHD that are being presented.

### **Mother of Samantha C.**

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My daughter Samantha C. was diagnosed with early-onset FSHD when she was eight years old in 2017. Since birth we struggled for an explanation for some of Sam's symptoms. She had hearing loss and a complete lack of expression in her face. She couldn't raise her eyebrows, she couldn't smile, and her speech was unintelligible. We were convinced that she must have nerve damage in her face which caused the lack of facial movement. We went to NYU and several ENT pediatric specialists in NY and NJ and they all said she just had low muscle tone. This went on for years. Finally, in 2017 at the age of 8 we went to CHOP. Within five minutes of meeting with Sam they said the words FSHD ... a progressive myopathy. Then they started checking her arms and legs for signs of muscle wasting... her arms and legs? The room spun and I almost fainted. I threw up several times outside the hospital that day. How could my seven-year-old, who runs, plays and climbs in the park, have a muscle-wasting condition that could put her in a wheelchair? I spent the next year in a "diagnosis hole," connecting with others affected and seeing the progression of this disease as it happened to people in their 20s and 30s, knowing this would happen to my baby. I cannot stress enough I would give my own life to get a treatment for this disease.

Since 2017 Sam has had progression. She can barely run anymore. She can't lift both arms overhead. She struggles to walk up the stairs.

Please, please help us! Everything I have read about FSHD proves it is unpredictable and variable. I can attest to the fact that one day Sam was fine and the next her shoulder was slouched and had lost muscle ... overnight! If there is a drug candidate on your table that does no harm and can potentially help, we NEED it. Time is not on our side. My daughter is not disabled yet. She is 11. Help her. Help us!

Please, please, help our children have a fighting chance of retaining the muscles they still have.



## Susie K.

July 1, 2020

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First day of the month! Well, another typical day with FSHD, meaning it took 45 minutes to get dressed, wash my face, and brush my teeth. I did practice my spiritual reading, so that was a positive! But still need to do my visualization and chair yoga.... It's now almost 4:30 and the bed is finally made, meaning I finally put the sheets on after sleeping without for five days. That simple little process, and I don't even put on a top sheet anymore, was over two-and-a-half hours. After both corners came off at the same time, I simply fell on a partially made bed and sobbed for half an hour. I'm not usually this fixated with time, but now I see where my day goes. Am I feeling sorry for myself or am I depressed? Or could it be this is becoming more difficult with each passing day? What will go next? I did realize this morning as I'm trying to whip up a protein drink, that the blender I have will not work any longer for me. Just too damn heavy to pick up, let alone pour into a glass!

I'm writing this as I sit propped up on my freshly made bed. I reach back for my water and ... you can't do that! You give it a couple more times trying to turn around or put my arms behind me to lift the glass and a sharp burning pain runs up my biceps ... are those going, too? Please no, as I depend on these to get me up, turn me over and so many other basic routines of life. Every day brings me to a new way of compensating the simplest of tasks. This new life is all about compensation.

I was diagnosed in June 2014 at the age of 63. I will be 69 in December. I had been having various symptoms, but I had no idea this disease even existed. I knew something was going on with my body, and I knew something wasn't right. My gait had changed dramatically over the three years prior. Stairs terrified me and at the time I lived in a home where everywhere you went stairs were involved. For me to get up and have my morning cup of coffee, it was three stairs down to the main floor and then 13 up to the kitchen. If I wanted to go to my office or my closet it was another 10. A gorgeous home, and originally we thought the stairs would keep us young. In theory, a great idea, but not for someone with a genetic disease she didn't know was in her family.

I started falling, and that was persistently a nuisance, as well as embarrassing. I can only remember one black eye, maybe two, but feel blessed, as it could've been much worse. Again, very hard to admit you fall for no reason. I lost weight (muscle) and I hovered around 88 for at least five years. Again, what was this all about? Stomach issues, gall bladder removed in 2012, and the same digestive issues remained. At times, I had difficulty eating, which was a total perplex! It was as though my mouth didn't have the strength to chew properly, or at times it was a challenge getting a utensil up to my mouth. I became very self-conscious. Food at times would simply fall out of my mouth, and for a woman that taught social manners for a living, this was beyond embarrassment.



## **Fourth of July, 2020**

Freedom Day! Doesn't really feel like one today, but I'll get to that later ... last couple of days were such an improvement over what I was going through three days ago. As I've said, every day is different and that means brings new challenges. Sometimes simple, but others are life changing. Realizing how much time it takes to embark on any kind of adventure, even if its simply taking a ride, has to be taken into consideration and then you need to count forward to determine your ETA! Or, have I got that backwards? The next couple of weeks are "field trips" to doctors, dentist, maintenance and God only knows what else. So many of these should've been months ago but, guess where we've all been?! The world of COVID and quarantine.

This week was the eye doctor to discuss cataract surgery. I live on an island where the only way to get here is by ferry. I bring this up as there is only one doctor on Whidbey Island that does these surgeries, making it so much easier for follow-ups. Every time you go to the other side, you've lost an entire day and oodles of energy in the process. So that literally ate up my day and the appointment was not even four miles away. I made up for it yesterday by never getting out of my pajamas, hair pulled back in a turban, maybe washed my face and brushed my teeth by 3:00? I did art the entire morning and it was such a delight ... watercolors, drawing, coloring. The simple visual of looking at my vast array of these colorful tools surrounding me brought such a simple joy.

## **July 6, 2020**

Today is a challenging day of a different sort! Staying in the moment and being mindful is the task at hand. One of my coping skills is to use this practice as often as I'm able. Pain vanishes, where in this realm it doesn't exist. My mantra is "I am not this body! My true essence is greater than this physical world of FSHD." Along with practicing the art of being mindful, I use Corey Hess, my structural integrator, for body work. Being in this stay-at-home mode of existence, all hands-on therapies have been cancelled. The last time Corey came to the house to do body work was at the beginning of February. Since then we've been doing distance sessions.

The sessions feel surprisingly palpable, tangible and objective to me. I am not making it up or trying to make something happen. I am dialoguing with the person's body/system/energy in the room with me in a very real way. When I'm doing this work, I feel the client's body respond and change. And, I feel myself change, my own body will fill with energy, as I interact with their system. The format for distance sessions is simple. The client and I agree on a time or the session to occur. I write them a quick message to let them know I am beginning. All the client is asked to do is hang out in a quiet, comfortable space while the session is happening. It lasts about half an hour. Immediately after the session, I write the client an email telling them what I found and felt during the session. Then, the client writes back with their personal feedback.



My personal response has been, "I feel cleared in my energy, very rested. My physical form seems more fluid and easy! Yes, my body was locked, but I feel as though I can maneuver in a way that actually feels good and grounded. You nailed it, Merlin!"

### **July 19, 2020**

Sitting on my deck on a perfect summer afternoon, gazing at the blue waters of Puget Sound, I forget what's happening with my body until I try to get up. At least five tries to push myself up just to catch the handles of my outdoor scooter. I jump on and ride to the sliding glass door and then transfer to my indoor scooter. It is the lack of my left side working that makes walking a thing of the past.

But today, my physical body feels good. No upset stomach or nausea. No pain. I am in a state of grace as I reflect on my past and the progression this disease has manifested. I truly never thought I would get this bad! Why did I ever think that? Who did I think I was to be spared this deterioration? I remember shortly after I was diagnosed, having a session with my intuit. She point-blank asked, "What is your biggest fear?" That caught me with a cold blank stare, until I responded, "Well, I won't be able to walk!" Her prompt response was, "So, what's the big deal about walking?" "Because everyone does!"

And now five years later, I cannot walk!

Could we just find something to halt the progression any further?

### **July 26, 2020**

Americans with Disabilities Act is 30 years old today....

Wow, I never gave this much thought 30 years ago. Looking back, I was in great physical and mental shape. I was happily married with two beautiful sons and living a life full of vim, vigor and energy. I was a national speaker and trainer who had written a book about manners. I had no idea this was looming in my future.

I am convinced my mother had FSHD, but she didn't know it. She died of pneumonia at the age of 52. Her signs were very similar to mine. Her gait was way off, falling constantly, the lordosis tummy, rounded shoulders, and she continually lost weight until she passed away. She was shamed by her community. Everyone blamed this on her drinking!

### **July 31, 2020**

Since this is the final day for me to submit my story, it's time to wrap this up. Hopefully, I've given you a small glimpse into my life and what living with FSHD is like for me. I'm frightened of what's



next ... will I be able to stay in my wonderful little home? Will I need a caretaker, and who would that be? Will I have enough money to take care of all of this and more? So many questions and very little answers at this point.

Fast track what ever you can for all of us suffering with a disease that we did not ask for. We will find a cure by 2025, if not before. But we need your help ... PLEASE!!

## Theresa F.

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How FSHD has affected my life

For five years I went from doctor to doctor before getting a definitive DNA diagnosis from the doctors at Johns Hopkins in 2010.

My first symptoms started when I had difficulty carrying something as light as a pizza box or walking up my driveway without a pulling sensation in the muscles that keep me upright.

I can no longer carry a newborn grandchild or lift another into a car seat. I can't go for a walk with my husband or climb stairs without using both hands to pull myself up by the rail. Most often I crawl up the stairs. I'm embarrassed to go to a mall because people often ask me, "Are you okay?"

I can no longer move a pot of water from the sink to the stove to boil water for pasta or put anything in the oven that would require two hands to hold it. If I try to put something in the microwave, I can only use one hand because I need the other to hold on to something to keep me upright while I lift the dish.

I'm afraid to take a shower without having the phone within reach because I don't feel steady on my feet.

Most of the time I have pain in the back of my right shoulder or when I try to lift my arms.

I often have difficulty swallowing food and getting enough volume in my voice for people to hear me.

I am no longer able to stand for any length of time before I have to sit down. I get so tired during the day I have to talk myself into moving.

Since that time, 10 years ago, I am constantly looking for signs of FSHD in my grandchildren and children who have a 50/50 chance of getting this disease. My husband and I decided not to tell our family of my diagnosis since there are still no drugs or treatment to even slow down the progression of FSHD. And I have become more disabled each day.



## Trisha G., Las Vegas, Nevada

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FSHD deserves no sugar coating. From the moment you notice that you've had problems with weakness in your arms and that gallon of milk for some reason has been getting harder to lift and carry. You've also become more easily fatigued walking or running for as fast or for as long as you once used to. Perhaps it's stress, or maybe you're just getting out of shape, you say. But it's been months now, a year, years even, and you just can't seem to turn it around.

Your shoulders and/or hips are always sore, you seem to trip all the time now. Your shoulders are droopy, your biceps have shrunk. A lot. You are worried and suspect something's off, but not too much, because the doctor is going to tell you it's just a hormonal imbalance or that you need to improve your diet, or something. It can't be anything that serious. The doc will be able to throw you some pills or tell you to eat more broccoli and you'll be back to your old self in no time.

So you go to your appointment, you tell them your symptoms. They run some tests, you squeeze their fingers and try to raise your arms. When did that get hard to do? They run their hands over your shoulders and down your spine. It tickles a little, but once you stop giggling, they whack you on the knees and you are asked to walk the hallway for them. The doctor leaves, comes back, after forever, with a pamphlet in hand, an FSHD diagnosis, and a referral with a "I'm sorry" and "have a nice day." What just happened?

You were told what it meant, maybe you have a family history of FSHD, but even so, the end of the appointment and the car ride home were a blur. The shock wears off sometime during the next meal and you begin to talk about it with your loved ones. What sticks out are words like muscle weakness, progressive, and possible wheelchair at some point in the future. Someone makes a crack about calling you "Wheels," you throw a look, finish your peas, and then you cry yourself to sleep that night.

Going forward, you google, you research, you join groups. Time marches forward, and so does your progression. You learn intimately that FSHD is not shy about living up to its hype.

Your arms become like pool noodles and it becomes too hard to carry your own groceries, raise a cup to your mouth, shampoo your own hair, wrap your own arms around someone to give a hug.

For the umpteenth time, you've been told to smile bigger, and you are, but it's only your eyes that get wider instead of your lips, so you feel like you look like a psycho killer. And then you hear the click of a camera.

The curve in your spine makes you look like an "s" and forces you to walk with a cane or walker, or with your hands in your back pockets, to stabilize your body, even if it's only for a few minutes, only





for a few feet, and causes excruciating pain. Forget dancing or moving fast at all with your wobbly legs, a feather could knock you over, you're just trying not to fall. At. All. Times.

You give up your driver's license because you have drop foot, and your arms and legs are not responsive enough to turn the steering wheel and use the pedals quickly and safely.

Your scooter or wheelchair becomes your legs.

It gets too hard to hold up an instrument and play.

You can no longer dress yourself, bathe yourself, use the toilet completely on your own.

FSHD just sucks the ability out of you.

## Steve and Darcie M.

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First, we would like to say thank you for soliciting statements related to early-onset FSHD. We're sorry we could not attend the live version of Voice of the Patient in June.

In our experience, FSHD is anything but a benign, adult disease; FSHD has affected our lives profoundly. We have four children, a boy and three girls. Our youngest daughters are fraternal twins who are now 26 years of age.

Our oldest, Robert, was born in 1988. We started noticing muscle weakness in Robert when he was a toddler. In retrospect, Robert had the trademark FSHD facial weakness much earlier. The symptoms progressed into the classic scapula winging and bicep weakening.

Our second child, Amanda, was born in 1990, and is not affected. When Robert was officially diagnosed with FSHD in 1994 at age six, Darcie was already pregnant with our twins.

Robert was diagnosed with infantile FSHD, relatively rare and much more severe; he was in a wheelchair permanently by age 12 with profound muscle weakness throughout his body. Now knowing what to look for, we noticed very similar symptoms in one of our twins. One twin was also diagnosed with FSHD as a child; her sister is completely unaffected by the disease.

Robert graduated high school in 2006 and completed college in early 2009 with a bachelor's degree in computer science. He worked in Salt Lake City and moved back to Idaho Falls to live with us in 2010. In 2012 Robert started to weaken even more, and due to lung issues almost died later that year. He was on oxygen and we placed him on hospice, weighing in at 66 pounds. Even though Robert made a remarkable recovery, "graduating" from hospice, he required full-time support from us. We made the decision to take care of Robert's needs for the rest of his life.



I do not know if I would call it a burden, but FSHD places a tremendously heavy emotional and physical load on the patient and caregiver(s), not to mention the patient's family. Due to the severe muscle weakness of an infantile FSHD patient, almost all daily activities must be taken care of. Dressing, bathing, cooking, feeding, toileting, and transferring all fall to the caregiver. The patient's siblings also watch as their beloved brother or sister slowly loses their ability to smile, walk, eat and even breathe.

In early 2017, Robert contracted a common cold and died a week later at the age of 29 due to effects of pneumonia. We chose to take care of Robert to the end, engaging hospice again for the last week of his life. Caring for a terminally ill child places an enormous load on the entire family of those most severely affected by this disease. The grief process never ends with the continual progression of the disease, and the associated adjustments to care that we just could not always foresee.

When I say our children are not affected, of course I mean physically. Siblings have an amazing bond that even us as parents do not fully grasp. Our surviving children and our entire family have been forever changed by this experience. Although we would most likely not choose this road if given the opportunity at the outset, we have grown immensely in this now almost 30-year journey.

Kendra, our twin daughter affected by FSHD, has less muscle wasting than her brother Robert, but still has significant weakness throughout her body and deals daily with the debilitating effects of the disease. She wears a brace on her left leg to compensate for foot drop. She has endured two scapulothoracic surgeries which are highly invasive and involve a painful recovery process but have aided in an increased range of upward motion of her arms. Kendra is currently participating in the ReDUX4 clinical trial.

It is too late for Robert, but we along with our daughter Kendra will always hold out hope for drugs and treatments that would slow down or even halt this terrible disease not just for our family, but for all the families who deal daily with this disease. To reverse the effects of FSHD and actually build muscle would be considered a miracle.

Thank you for the opportunity to be part of the Voice of the Patient.



## ANONYMOUS: Written testimony

AGE	DATE	MILESTONE	DESCRIPTION
	8/1957	Born in Chicago.	
18	9/1975	Diagnosed with FSHD around a year after weakness first appeared. It was my first month of college, and my first time living away from home. Diagnosis made by Dr. Michael Brooke, Washington University Medical School.	Genetic test didn't exist. Diagnosis was based on clinical observation of symptoms, electromyograms, muscle biopsies, CPK level, and other blood tests.
18	1975	The last time I rode a bicycle.	I tried to ride a bike the summer after I was diagnosed, but didn't have enough stability.
21	12/1978	Had scapular fusion surgery on my left shoulder performed by the surgeon who developed the procedure. I was around the ninth person to have it. It improved and prolonged my ability to lift my left arm, but there were major side effects.	Lifelong side effects from three months in a heavy, confining body cast: accelerated and increased weakness in trunk, abdominal and upper leg muscles. Also, the surgeon fused my shoulder too high, resulting in asymmetry and reduced range of motion, which make certain movements and activities of daily living much more difficult.
24/25	1982	Graduated from law school in June and began practicing law in San Francisco in October or November.	One of the primary reasons I moved to San Francisco after law school was to live somewhere without snow. I had fallen in the snow and ice in Chicago, St. Louis and Boston, and it would be safer and easier to live where it never snowed.
25	10/1982	The last time I was able to get out of a bathtub.	I haven't taken a bath since October 1982.
32	Spring 1990	Began using a wheelchair. Walking had become increasingly difficult and precarious. Falls were getting worse. I had been in some denial about my loss of ability, and I had been afraid to begin using a mobility device.	By the end of 1990 I was using a power wheelchair all of the time outdoors and most of the time indoors.
33	1990	Acquired a lowered floor, wheelchair accessible minivan.	I was able to drive without hand controls. The driver's seat moved along a rail, and I transferred from my wheelchair to the driver's seat.



AGE	DATE	MILESTONE	DESCRIPTION
33	1990	Three years before we were married, my wife and I, who lived together, saw a genetic counselor and decided not to have children.	We decided not to have children because: (1) my FSHD was relatively rapidly progressive, with all of the medical, practical, logistical, emotional, physical, financial and career difficulties and uncertainties my progression entailed for both of us; and (2) the probability of passing on FSHD to each child was 50%.
38	July 1996	Broke my left femur and lost the ability to stand or walk. Began using a power wheelchair 100% of the time.	This despite three weeks of intensive inpatient rehabilitation and several months of outpatient rehabilitation.
Early 40s	Late 1990s	Lost the ability to put on and remove a jacket or coat, due to my inflexible, elevated left shoulder and weak right shoulder.	
40 or 41	1998	Stopped driving.	My driving was evaluated at an adaptive driving program. The potential solutions were uncertain, complicated and expensive.
42	1999	Tested genetically positive for FSHD. I decided not to find out how many D4Z4 repeats I have. (Scientific consensus is that, combined with other factors, the fewer repeats the more severe the FSHD.)	I took the genetic test at the request of my sister when she was planning a family.
42 or 43	1999 or 2000	Began using a BiPAP every night, all night.	Respiratory insufficiency due to weak diaphragm muscles. This is not uncommon for people whose FSHD is severe and advanced.
44	Spring 2002	Stopped working and applied for disability. Began receiving Social Security Disability and payments under a group long-term disability insurance plan. After the requisite waiting period, I became covered by Medicare because I was on SSDI (this was after 2002).	I had practiced law for 19½ years. Despite reasonable accommodations from my law firm, working had become too difficult because of my FSHD. I was increasingly fatigued and decreasingly productive. My FSHD was progressing more quickly.
44	2002	Began using an electric bed.	The foot of the bed and head of bed can be separately raised. Entire bed can be elevated.



AGE	DATE	MILESTONE	DESCRIPTION
44/45	2002	Lost the ability to transfer from wheelchair to toilet without assistance. Shortly thereafter I lost the ability to transfer to toilet even with assistance.	
44/45	2002	Began using a shower/commode chair. I was able to transfer from wheelchair to shower/commode chair using a transfer board and with a great deal of assistance.	
44/45	2002	The last time I used a public toilet. Since that time I've only used the toilet at my home, at hotels, and at my parents' house, always with a shower/commode chair and with a great deal of assistance.	
48 or 49	2005 or 2006	Began hiring caregivers several mornings per week at home to assist me with toileting, showering, dressing, and other activities of daily living. Over subsequent years I've had caregivers most mornings each week, and for more hours.	Until 2005 or 2006, my wife had been helping me with ADLs without a respite.
52	2009	Began hiring caregivers when traveling to assist me with toileting, showering, dressing, and other activities of daily living.	Until that time, when we traveled, my wife had been helping me with ADLs without a respite.
53	2010	Lost the ability to transfer from bed to shower/commode chair, and vice versa, which I had been able to do by using a transfer board and with a great deal of assistance. Installed a ceiling lift in the bedroom, which my wife and caregivers use to transfer me from bed to shower/commode chair, and vice versa.	The ceiling lift is not operable independently. The lift wasn't covered by insurance.
56	2013	Got a Cough Assist device.	On standby, used if I have a bad cold.
58	2015 (approx.)	Eating and drinking became increasingly difficult. The type of food, design of silverware, type of table, and other factors affected my ability to eat and drink. Around this time I began drinking with a straw.	Over the years I developed complex maneuvers for eating and drinking - leaning to my right, using momentum to swing and elevate my left arm, and supporting it with my right arm.



AGE	DATE	MILESTONE	DESCRIPTION
59 or 60	2016 or 2017	Lost the ability to transfer from bed to wheelchair, which I had been able to do by elevating the bed, using a transfer board, and with a great deal of assistance. I was still able to transfer from wheelchair to bed by elevating the wheelchair, using a transfer board, and with a great deal of assistance.	It's more difficult to transfer from a larger surface, such as a bed, to a smaller surface, such as a wheelchair, than to transfer from smaller to larger.
60	9/2017	My last airplane flight.	In 2018, I decided to stop traveling by air because it had become too difficult and fatiguing.
60	12/2017	Venous wound in my leg. This was my first, and so far my only, venous wound.	Began wearing compression stockings daily and most nights.
60/61	2018	Since around early 2018, I haven't been able to feed myself at a regular height table at all; I need to be fed.	However, as of early 2020, with my wheelchair fully raised, I'm still able to feed myself at a high counter if conditions are right (height of counter, depth of overhang, type of food, type of silverware), but even then, I need some help.
61	11/2018	A spirometry test measured my forced vital capacity at 40% of normal. Acquired a Trilogy 100 for occasional mouthpiece ventilation during the day.	
61	6/2019	Lost the ability to transfer from wheelchair to bed, which I had been able to do by elevating the wheelchair, using a transfer board, and with a great deal of assistance. This was the last of the four transfers that I had been able to do.	At this point my wife and caregivers use the ceiling lift to transfer me for all transfers. It's extremely difficult to travel overnight, even locally, because without a ceiling lift, it takes two people to lift me for all transfers. I'm 6 feet tall and 190 pounds.
62	Early 2020	My hands and fingers have been weak for years.	I haven't been able to pick up coins or small paperclips, or use a stapler, for several years. It's become difficult to fold currency and put it in my wallet. I can't open most bottles or jars. I can't open mail unless the envelope is loosely sealed.



A meaningful illustration of the degree of my disability is the equipment that I use. Here's my equipment:

- A manual wheelchair, which was my first wheelchair and which I acquired before I got a power wheelchair. Sometimes friends have used it to lift me up stairs into their homes for social gatherings, but this has become increasingly difficult as we've gotten older, so we haven't done it for several years. I keep this chair for evacuation in an emergency.
- Two power wheelchairs (my primary chair and a backup chair)
- Several wheelchair seat cushions and cushion covers
- Some chest belts
- Two wheelchair battery chargers (when I traveled to Europe, I also had one with 110/220 Volt dual settings)
- Laminated instructions (when I traveled to Europe, I also had instructions in French and Italian) for the airlines on how to handle my power wheelchair
- A commode/shower chair (and various backrests, seat cushions and spare parts)
- A custom-designed, custom-fabricated removable grab bar for the bathroom
- A shower bench
- A standing frame
- A reading stand
- A ceiling lift
- An electric bed
- A residential elevator and electric door opener
- A lifeline emergency alert system
- A BiPAP and a backup BiPAP
- Two battery backups for the BiPAPs (purchased in 2019 in preparation for a possible public safety electrical outage during California fire season)
- A Cough Assist machine
- A Trilogy ventilation device
- Various accessories for the respiratory devices (tubing, filters, interfaces)
- Adaptive equipment for my computer (large trackball, tabletop microphone, voice recognition software before it became standard)
- A lowered floor, wheelchair accessible minivan
- Assorted tools, accessories and spare parts

Acquiring, managing, documenting, maintaining and repairing this equipment takes time, energy, money, organizational skills, and help from my wife and my caregivers. The equipment takes up space in our home. Before acquiring something, I research it and usually try it. Each item must be maintained and repaired. There are a small number of medical equipment dealers in and around





San Francisco, they are busy, and it can be difficult to schedule a repair. I document repairs in detail, which helps in troubleshooting if the same problem arises again. Fortunately, some of my caregivers are mechanically skilled, and they do small repairs and adjustments. Equipment needs to be replaced when it wears out or breaks beyond repair, and when replacement parts are no longer available. I also replace equipment when it is no longer effective for me because my needs have changed as my condition has progressed, and when a better technology or product becomes available if it better suits my needs.

Each item must be paid for. Items covered by insurance require a doctor's prescription or sometimes a detailed letter. Insurance doesn't cover everything. The ceiling lift, for example, wasn't covered by insurance and, what's more, was subject to California sales tax because it wasn't on the list of tax-exempt medical items. Medicare doesn't pay for the seat-elevating feature, lights, and certain other components of power wheelchairs. For each item that isn't fully covered by insurance, I keep detailed cost records for tax purposes.



## APPENDIX IV: Live-polling questions

### TOPIC 1: About you

#### 1. Are you (check all that apply):

- a. An individual living with FSHD
- b. A parent or caregiver of an individual with FSHD

#### 2. Where do you live?

- a. US: Northeast
- b. US: Mid-Atlantic
- c. US: Midwest
- d. US: South
- e. US: Mountain
- f. US: West
- g. Canada
- h. Central or South America
- i. Europe or UK
- j. Asia
- k. Africa
- l. Australia or New Zealand
- m. Other

#### 3. What is your age?

- a. Younger than 18
- b. 18–29 years
- c. 30–39 years
- d. 40–49 years
- e. 50–59 years
- f. 60 or older

**4. How do you identify?**

- a. Male
- b. Female
- c. Gender variant/non-conforming
- d. Prefer not to answer

**5. How long ago were you diagnosed with FSHD?**

- a. Within the past 5 years
- b. Between 5 and 10 years ago
- c. More than 10 years ago
- d. Not sure

**6. Have you had a genetic diagnosis and if so, what was the result?**

- a. Type 1
- b. Type 2
- c. Both Type 1 and 2
- d. Results were “inconclusive”
- e. Not sure
- f. I was never tested

**TOPIC 2: Disease symptoms and daily impacts that matter most to patients****1. How difficult are activities involving your upper extremities?**

- a. Not at all
- b. Mildly
- c. Moderately
- d. Severely

**2. How difficult are activities involving your core & lower extremities?**

- a. Not at all
- b. Mildly
- c. Moderately
- d. Severely



**3. Of all the symptoms you have experienced because of FSHD, which have the most significant impact on your daily life? Select up to three.**

- a. Impaired mobility
- b. Pain
- c. Fatigue
- d. Impaired facial expression
- e. Depression or anxiety
- f. Difficulty using hands or arms
- g. Poor sleep
- h. Speech and/or swallow difficulties
- i. Breathing issues
- j. Hearing impairment
- k. Impaired vision
- l. Urinary or bowel incontinence
- m. Other

**4. Which every day activities are important to you that you cannot do at all or as fully as you would like because of your condition? Select up to three.**

- a. Walking
- b. Attending school or working
- c. Personal hygiene
- d. Being intimate with a spouse or partner
- e. Driving a motor vehicle
- f. Participating in sports or hobbies
- g. Performing household tasks
- h. Participating in family care and activities
- i. Going out, socializing, traveling
- j. Other



**5. What worries you most about your condition in the future? Select up to three.**

- a. The stress of not knowing how the disease will progress
- b. Losing independence
- c. Losing mobility/ability to walk
- d. Losing ability to communicate and/or swallow
- e. Losing ability to breathe and developing respiratory issues
- f. Not having the energy to work and live as I want to
- g. Having to cope with pain
- h. Not knowing if I can support myself/family financially
- i. Becoming a burden to my family
- j. Losing social connections
- k. Other

**TOPIC 3: Current approaches to managing the condition**

**1. Are you using any of the following to manage FSHD symptoms? Select all that apply.**

- a. Prescription medications (such as pain relief, anti-depression/anxiety, steroids)
- b. Over-the-counter medications (such as acetaminophen, ibuprofen)
- c. Medical or recreational marijuana, cannabidiol (CBD)
- d. Dietary and herbal supplements
- e. Not currently using any

**2. Beyond medications and supplements, are you using any of the following to manage FSHD symptoms? Select all that apply.**

- a. Exercise
- b. Physical or occupational therapy
- c. Braces, Kinesio tape, etc.
- d. Mobility aids (such as walker, scooter, wheelchair)
- e. Surgery (such as scapular fixation)
- f. Diet modifications
- g. Complementary or alternative therapies
- h. Counseling/therapy
- i. Other
- j. Not currently using any

**3. How well does your current regimen control your condition overall?**

- a. Not at all
- b. Very little
- c. Somewhat
- d. To a great extent
- e. Not applicable because I'm not using anything

**4. What are the biggest drawbacks of your current approaches? Select up to three.**

- a. Not very effective
- b. High cost or co-pay, not covered by insurance
- c. Limited availability or accessibility
- d. Number of pills/medications needed per day
- e. Side effects
- f. Requires too much effort and/or time commitment
- g. Other
- h. Not applicable as I am not using any treatments

**5. Short of a cure, what outcome is the most meaningful to you in a future treatment? Select your top choice.**

- a. Slowing or stopping the loss of muscle function
- b. Regaining strength and/or muscle function
- c. Lessening pain or fatigue
- d. Preserving respiratory and lung function
- e. Improving hearing/sight loss
- f. Other



## APPENDIX V: Pre-meeting survey questions

**Directions.** This survey is to be answered by or about a single individual who has been diagnosed or told they have FSH muscular dystrophy. (A parent, guardian, or caregiver can respond on behalf of an affected individual. Please answer the questions as if you are the patient.) Responses are tied to a specific email. If you have other family members who also have FSHD, please make sure they are each sent the survey to their own email address. Your responses are confidential.

### SURVEY 1: About you

#### 1. Are you (check all that apply):

- a. An individual living with FSHD
- b. A parent or legal guardian of a child with FSHD
- c. A representative of an adult with FSHD

#### 2. Where do you live?

- a. United States [enter state]
- b. Canada
- c. Central or South America [what country?]
- d. Europe or UK [what country?]
- e. Asia [what country?]
- f. Africa [what country?]
- g. Australia or New Zealand
- h. Other [what country?]

#### 3. What is your (i.e., the patient's) age?

- a. Younger than 18
- b. 18-29 years
- c. 30-39 years
- d. 40-49 years
- e. 50-59 years
- f. 60 or older



**4. How do you (i.e., the patient) identify?**

- a. Male
- b. Female
- c. Other

**5. What is your (i.e., the patient's) race/ethnicity (check all that apply)?**

- a. White [text box: countries of origin]
- b. Black [text box: countries of origin]
- c. Hispanic [text box: countries of origin]
- d. Asian [text box: countries of origin]
- e. Near or Middle East [text box: countries of origin]
- f. Native American or Alaskan [text box: countries of origin]
- g. Pacific Islander [text box: countries of origin]
- h. Other

**6. What is your (i.e., the patient's) annual household income?**

- a. Under \$15,000
- b. \$15,000-\$24,999
- c. \$25,000-\$49,999
- d. \$50,000-\$74,999
- e. \$75,000-\$99,999
- f. \$100,000-\$149,999
- g. \$150,000-\$199,999
- h. \$200,000 and above

**7. Your (i.e., the patient's) current occupation is:**

- a. Student
- b. Employed outside home
- c. Self-employed
- d. Homemaker
- e. Unemployed
- f. On disability
- g. Retired
- h. Other

**8. How long ago were you diagnosed with FSHD?**

- a. Within the past 10 years
- b. More than 10 years ago
- c. Not sure

**9. Have you had a genetic diagnosis and if so, what was the result?**

- a. Type 1
- b. Type 2
- c. Both Type 1 and 2
- d. Not sure
- e. I was never tested

**SURVEY 2: Disease symptoms and daily impacts that matter most to patients****1. How severely are you affected? Check all that apply.**

- a. Facial weakness
- b. Mild scapular winging
- c. Can't raise arms above shoulder height
- d. Severe scapular winging
- e. Can't raise arms more than 60 degrees from the side of my body
- f. At least one muscle in my arm can lift against gravity but not against additional resistance
- g. Foot drop
- h. Unable to stand up from a chair unless supported on just one side
- i. Unable to stand up from a chair unless supported on BOTH sides
- j. I can walk unaided at all times but worry about stairs, fatigue, and falling
- k. I can walk but use a scooter or wheelchair for distances greater than ~100 yards
- l. Use a scooter or wheelchair virtually all the time
- m. Need a breathing assist (such as BiPAP) at night
- n. Need a breathing assist during the day
- o. Other

**2. Of all the symptoms you have experienced because of FSHD, what are the top three symptoms that you consider to have the most significant impact on your daily life?**



3. Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of your condition? List the three most important activities that have been impacted.

### **SURVEY 3: What worries you the most about the future?**

What worries you most about your condition? List your top three concerns.

### **SURVEY 4: What are you doing now to manage your condition?**

1. Are there specific symptoms that you are doing something to try to improve? Which symptoms?
2. Have you ever used prescription medications to help with your symptoms? List all that you can recall.
3. What else are you doing to manage any of your FSHD symptoms? List your top three interventions.
4. What are the biggest downsides of your current approach? List up to three.

### **SURVEY 5: What are you looking for in a better treatment?**

1. What outcomes are most meaningful to you in a future treatment?
2. Short of a complete cure for your FSHD, what would be the most important benefit that a treatment could provide?