

Early-Onset Facioscapulohumeral Muscular Dystrophy (FSHD)

Patient Listening Session Summary

Every Age. Every Stage. A Cure For All.

Prepared by:

FSHD Cure Initiative

In partnership with FSHD Society and Solve FSHD

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I. About the FSHD Cure Initiative

FSHD Cure Initiative is a nonprofit organization dedicated to accelerating the development of and access to effective treatments for all individuals affected by Facioscapulohumeral Muscular Dystrophy (FSHD), with a particular emphasis on those with early-onset disease. Guided by its mission, “Every Age. Every Stage. A Cure For All,” the organization works closely with patients, families, clinicians, researchers, industry, foundations, and regulatory agencies to advance therapeutic development for individuals affected by FSHD across every age and stage of disease. Recognizing that individuals with early-onset FSHD experience some of the greatest disease burden yet have historically had limited opportunities to participate in clinical research, FSHD Cure Initiative is committed to ensuring that their experiences inform therapeutic development and regulatory decision-making.

II. Disease Background

FSHD is a rare, relentlessly progressive, autosomal dominant muscular dystrophy affecting approximately 1 in 8,000 individuals worldwide, however there are only 7,000-9,200 with a known medical encounter with FSHD identified in the United States. The disease is caused by contraction of the D4Z4 repeat region on chromosome 4q35, resulting in inappropriate expression of the toxic DUX4 protein, which leads to progressive skeletal muscle degeneration. Two genetic subtypes—FSHD1 (approximately 95% of cases) and FSHD2—are clinically indistinguishable and share the same underlying disease mechanism of pathogenic DUX4 expression.

FSHD is characterized by progressive weakness of the facial, scapular, upper extremity, trunk, and lower extremity muscles, accompanied by a broad spectrum of extra-muscular manifestations. Patients may experience hearing loss, retinal disease, spinal deformities (including scoliosis and lordosis), respiratory impairment, chronic pain and fatigue, swallowing difficulties, cardiac abnormalities, and orthopedic complications. Disease progression is highly variable, but muscle weakness is irreversible and ultimately affects nearly all skeletal muscle groups.

Early-onset FSHD represents the most severe end of the disease spectrum and is typically associated with more rapid progression, earlier loss of ambulation, greater functional impairment, more frequent extra-muscular complications, and substantially higher mortality than classical adult-onset disease. Although clinical severity differs, early-onset and adult-onset FSHD are driven by the same underlying molecular disease mechanism. Childhood-onset cases represent approximately 20% of the overall FSHD population and early-onset (facial weakness prior to 5 years of age) represent about 10% of the FSHD cases. However, 55% of adult-diagnosed FSHD individuals reported having symptoms prior to age 18, regardless of when they were diagnosed. There are currently no FDA-approved disease-modifying therapies for FSHD. Clinical management is limited to supportive interventions, including physical and occupational therapy, assistive devices, orthopedic surgery, respiratory support, and symptom management, that do not alter the underlying disease course. Although multiple investigational therapies targeting DUX4 are in clinical development, children and individuals with advanced disease have historically had limited opportunities to participate because of age restrictions and disease severity-related trial eligibility criteria. Current scientific understanding suggests that therapies targeting the underlying disease mechanism have the potential to benefit individuals across the full spectrum of FSHD,

including pediatric patients and those with advanced disease, by preserving muscle, function, independence, and quality of life.

III. Patient Listening Session Overview

Patient Listening Sessions are informal meetings that give patients, caregivers, and advocacy organizations the opportunity to share their firsthand experiences directly with FDA staff. These sessions help the Agency better understand the impact of living with a disease or condition, the challenges patients face, and the treatment outcomes that matter most to the community. By incorporating patient perspectives into the medical product development process, Patient Listening Sessions help ensure that the patient voice informs regulatory decision-making and future therapeutic innovation.

The FDA Patient Listening Session on Early-Onset FSHD was held on July 1, 2026, organized by the FSHD Cure Initiative in partnership with the FSHD Society, Solve FSHD, and the FDA Public Engagement Staff. The 90-minute session featured patient and caregiver perspectives from seven panelists representing the full continuum of the early-onset FSHD experience—from childhood through young adulthood, including both patients living with FSHD and parents who have witnessed its devastating progression in their children. This Patient Listening Session built on previous FSHD patient community engagements with the FDA, including the July 2020 Externally-Led Patient-Focused Drug Development meeting and the August 2024 Patient Listening Session on Upper Body Mobility (reports available [here](#) and [here](#)); however, this was the first and only meeting to focus on the unique experiences and needs of the Early-Onset FSHD population.

IV. Objectives of the Session

The Early-Onset FSHD Patient Listening Session was convened by the FSHD Cure Initiative and its partner organizations to provide the FDA and other regulators with firsthand accounts of the lived experience of early-onset FSHD. The session sought to:

- Increase FDA understanding of the clinical manifestations, disease progression, and daily burden associated with early-onset FSHD across the lifespan, including its impact on childhood, adolescence, and the transition to adulthood.
- Illustrate the profound physical, emotional, social, educational, and caregiving impacts of early-onset FSHD on patients and families.
- Describe the current treatment landscape and the substantial unmet medical need for disease-modifying therapies capable of preserving function, slowing progression, and improving quality of life.
- Describe patient perspectives regarding participation in clinical research, including willingness to accept risk and barriers to enrollment.
- Communicate the urgent need for clinical trials that include pediatric and non-ambulatory patients—the populations with the greatest unmet need who are currently excluded from most ongoing research;
- Communicate which treatment outcomes patients consider most meaningful, including preservation of mobility, upper limb function, respiratory function, independence, and participation in everyday life.

- Reinforce that early-onset FSHD is part of the broader FSHD disease continuum and that therapies targeting the underlying disease mechanism should also be developed for individuals across all ages and stages of disease.

V. Attendance

The Early-Onset FSHD Patient Listening Session brought together representatives from the FDA, federal research organizations, and the FSHD community to foster meaningful dialogue about the lived experience of this rare disease.

The session included FDA staff from 19 offices and divisions across four FDA centers and offices. Attendees represented expertise in rare diseases, pediatrics, neuroscience, biologics, devices, orphan products, and pediatric therapeutics, reflecting broad Agency interest in understanding the needs of the early-onset FSHD community.

Office of the Commissioner (OC)

- Office of External Affairs/Public Engagement Staff (organizer)
- Office of Pediatric Therapeutics
- Office of Orphan Products Development

Center for Biologics Evaluation and Research (CBER)

- Office of the Center Director
- Office of Therapeutic Products/Policy and Special Projects Staff
- Office of Clinical Evaluation/Division of Clinical Evaluation General Medicine/General Medicine Branch II
- Office of Clinical Evaluation/Division of Clinical Evaluation General Medicine/General Medicine Branch III

Center for Drug Evaluation and Research (CDER)

- Office of the Center Director
- Office of New Drugs
- Office of New Drugs/Office of Neuroscience/Division of Neuroscience I
- Office of New Drugs/Office of Neuroscience/Division of Neuroscience II
- Office of New Drugs/Office of Rare Diseases, Pediatrics, Urology and Reproductive Medicine/Division of Rare Diseases and Medical Genetics
- Office of Translational Science/Office of Biostatistics/Division of Biometrics I

Center for Devices and Radiological Health (CDRH)

- Office of Product Evaluation and Quality (OPEQ)/Office of Health Technology I/ Division of Health Technology I
- OPEQ/Office of Health Technology III/ Division of Health Technology III
- OPEQ/Office of Health Technology III B/ Division of Health Technology III B
- OPEQ/Office of Health Technology V/ Division of Health Technology V B
- OPEQ/Office of Health Technology VIII/ Division of Health Technology VIII C
- Office of Strategic Partnership and Technology Innovation/Office of Equity and Innovative Development/Division of Patient Centered Development

Additional federal participants included representatives from the Reagan Udall Foundation and the National Institutes of Health's National Center for Advancing Translational Sciences (NCATS).

Representing the FSHD community were seven patient and caregiver panelists, organizational leaders, and a clinician with experience treating FSHD patients:

- Sara, age 17, San Diego, CA — Patient, diagnosed with FSHD at age 7
- Sam, age 20, Tucson, AZ — Patient, diagnosed with FSHD at age 6
- Maggie, age 17, Ann Arbor, MI — Patient, diagnosed with FSHD at age 13
- Vera, Bayonne, NJ — Caregiver/Patient, mother of Isla (10) and Siena (8), both diagnosed as toddlers; Vera herself diagnosed at age 35
- Justin, age 35, New Haven, CT — Patient, diagnosed with FSHD at age 4
- Kate, age 15, Boston, MA — Patient, diagnosed with FSHD at age 6
- Angela Farkas, Nashville, TN — Caregiver, mother of Alyssa (diagnosed at age 6; passed away at age 16) and Nessa, age 17 (diagnosed at age 2)
- Ally Roets, FSHD Cure Initiative — Parent
- Anna Gilmore, FSHD Society
- Eva Chin, Solve FSHD
- Dr. Russel Butterfield, MD, PhD — Pediatric Neurologist, University of Utah (clinician perspective)

Collectively, the panel represented the full continuum of early-onset FSHD, including children, adolescents, young adults, adults, ambulatory and non-ambulatory individuals, and a family who had lost a loved one to the disease. Their experiences illustrated the lifelong progression of early-onset FSHD and its profound physical, emotional, and caregiving impact, providing FDA staff with insight into the evolving burden of disease across the lifespan.

James Valentine, JD, MHS and Sarah Wicks, JD, MPH of Hyman, Phelps & McNamara, P.C. (HPM) helped prepare FSHD community participants and attended the session. James Valentine moderated the patient/caregiver roundtable discussion.

VI. Financial Disclosures

FSHD Cure Initiative and Solve FSHD do not receive funding from pharmaceutical sponsors. The FSHD Society receives funding from a variety of sources, including pharmaceutical sponsors developing treatments for FSHD. None of the funds used by the organization were used for the purpose of organizing or participating in this session. None of the participants in this meeting received compensation for attendance or participation.

Dr. Butterfield has received funding via contracts for research from Ionis Pharmaceuticals. He serves on scientific advisory boards for Sarepta Therapeutics, Biogen, Novartis, Precision Bio, Solid Bio, Bridge Bio, and Scholar Rock.

HPM received funding from FSHD Society for participation as consultants to help develop this session. HPM also receives funding from a variety of other sources, including sponsors, none of which were used for the purposes of organizing or participating in this session.

VII. Summary of the Topics Discussed

A. Early-onset FSHD is a severe, progressive form of the same disease affecting individuals across the FSHD spectrum.

Across all patient and caregiver presentations, participants described early-onset FSHD as the most severe end of the FSHD disease spectrum rather than a distinct disease. Although disease severity and age at onset varied considerably, speakers consistently emphasized that all individuals share the same underlying disease process and experience continual loss of muscle function throughout their lives. Participants described symptoms beginning in infancy or early childhood, including facial weakness, hearing loss, gait abnormalities, and delayed motor development, followed by progressive disability affecting nearly every aspect of daily life.

Vera, who was herself diagnosed with FSHD at age 35 while her daughters Isla (10) and Siena (8) were diagnosed as toddlers, embodied this spectrum within a single family. Vera described her own comparatively mild, later-recognized presentation—hearing loss, an asymmetrical smile, and reduced endurance she had long “*adapted and brushed aside*”—along her daughters’ severe, rapidly progressing early-onset disease, even though all three share the same genetic diagnosis.

Angela, mother to oldest daughter Allysa (16; passed away in 2021) and younger daughter Nessa (17), emphasized that even within the same family, disease progression can vary substantially while remaining equally devastating. She explained: “*Both of my daughters have experienced severe forms of FSHD, but in very different ways, with different timelines and different losses of function.*” Allysa’s disease progressed rapidly and severely—she was in a wheelchair by third grade, developed significant facial weakness affecting her speech, and ultimately required a feeding tube before passing away at age 16—while Nessa’s symptom’s developed more gradually, only becoming markedly limiting as she entered her teenage years, when her scoliosis and lordosis progressed to the point that she could walk only short distances and had to transition from in-person to online school.

Adults living with early-onset FSHD similarly reflected on symptoms beginning in infancy. Justin (35), who was diagnosed at age 4, described hearing impairment as his earliest manifestation before progressive weakness led to wheelchair dependence during adolescence and continuing disease progression into adulthood.

Although age of onset and severity varied considerably, no speaker described early-onset FSHD as a fundamentally different disease. Rather, they consistently characterized it as the most severe presentation along a continuous spectrum of FSHD driven by the same underlying molecular mechanism. Several participants noted that this irreversible disease progression continues throughout adulthood, reinforcing that early-onset FSHD is not simply a pediatric condition but a lifelong disorder with cumulative consequences. Collectively, participants emphasized that while early-onset disease often progresses more rapidly than classical adult-onset FSHD, it represents the same underlying disease and should be considered part of a continuous disease spectrum for drug development and treatment.

B. Early-onset FSHD is relentlessly progressive, stealing function, independence and childhood itself.

Across all patient and caregiver presentations, the defining characteristic of early-onset FSHD was its relentless progression. Participants consistently described a disease that does not plateau, but instead continually and irreversibly steals muscle function, independence, and opportunities throughout childhood and into adulthood. Rather than experiencing a single disabling event, patients and caregivers described an ongoing cycle of adaptation as new muscle groups weaken and everyday tasks that were once routine gradually became impossible. They emphasized that disease progression can be acutely rapid, frequently outpacing normal childhood development and experiences, and forcing children and families to continually adjust to new limitations.

Vera offered one of the starkest illustrations of this velocity: her oldest daughter Isla lost all mobility within 24 months of her first symptoms, and her younger daughter Siena stopped walking almost overnight: *“One day she walked into school. The next, she never walked again.”*

Kate (15) summarized this experience by stating, *“My childhood was measured not by milestones of growth, but by a timeline of losses.”* She described losing her smile, her ability to run, raise her arms, and ultimately walk independently before entering high school.

Maggie (17) echoed this sentiment, explaining that every year brought another major decline in function. *“Since my diagnosis, I have lost the ability to raise either arm, walk normally, or run at all. My loss happened in stages, with a huge new loss striking me every year... Pieces of me have slipped away and the activities I’m grateful to hold onto are next,”* she said, after describing the loss of her shoulder strength, core muscles, ankle function, and eventually the sports and family traditions she loved.

Several participants described the constant adaptations required to preserve independence. Maggie explained, *“Imagine moving around throughout the day with your arms tied to your waist so you can’t raise them above your chest. It’s very hard to function.”* Despite this, she described continually finding creative ways to compensate, using walls or momentum to move her arms and complete everyday tasks.

The loss of ambulation and upper-limb function was nearly universal and often strikingly rapid. Sam (20) developed foot drop by age 8, progressed to a walker by age 11, and was using a wheelchair full time by age 13; Kate stopped walking independent at age 9 and has since lost even the ability to lift her thumb for a full thumbs-up; and Vera’s daughters Isla and Siene each lost the ability to walk within months, leaving both dependent on others for nearly every physical task.

Participants repeatedly emphasized that losses extend far beyond mobility. Everyday activities such as dressing, bathing, eating, attending school, driving, working, participating in sports, and spending time with friends became progressively more difficult as muscle weakness advanced. Kate reflected that *“I have an independent mindset in a dependent body,”* describing the constant planning required simply to navigate school, social activities, and inaccessible environments.

When reflecting on their futures living with Early-Onset FSHD, participants described worries of how continued progression and further disability will eventually affect future independence. Sara (age 17) described the uncertainty experienced by many individuals living with FSHD, explaining,

“Every moment of weakness makes me fear for my future... Will I, in three years, be unable to feed myself?”

Others described how progressive disability affected future careers and financial independence. Sam explained that despite pursuing college and volunteering toward a career in audio engineering, he worries that continued muscle loss will make employment impossible. *“I worry about how much strength I will lose, and how I'll be able to provide for myself... I don't want to be a burden on the government or my family.”*

Parents similarly described watching independence disappear in real time. Vera explained that her daughters now depend on others for nearly every aspect of daily living and concluded, *“We are simply watching this disease take more from our children in real time.”* Angela similarly described watching her surviving daughter gradually lose the ability to attend school in person, spend time with friends, and participate in activities that other teenagers take for granted, noting *“watching these opportunities quietly disappear has been one of the most heartbreaking parts of this disease.”*

Across all presentations, participants described early-onset FSHD as a disease of continual loss. They emphasized that progression affects not only muscle strength but also independence, education, employment, relationships, and participation in the experiences that define childhood and young adulthood. Rather than fearing a distant future, many participants described living with the uncertainty of not knowing what ability would be lost next, underscoring the urgent need for therapies capable of preserving function before it is permanently lost.

C. Early-onset FSHD is a multi-system disease whose burden extends far beyond skeletal muscle weakness.

While FSHD is characterized as a muscular dystrophy, panelists made clear that its threat extends far beyond skeletal muscle weakness. Many described respiratory insufficiency, spinal deformity and orthopedic complications, hearing and vision problems, facial weakness, chronic pain, fatigue, and diminished bone density as significant and interconnected contributors to disease burden.

Respiratory decline was described as one of the most dangerous dimensions of the disease. Justin described the cumulative effects of advanced disease, including scoliosis severe enough to impair lung capacity, chronic respiratory failure requiring nighttime ventilation, osteoporosis, pressure injuries, tachycardia, and impaired vision. He recounted one instance of respiratory compromise that nearly cost him his life, *“Most concerning was an incident last summer where I almost died. Breathing issues caused a large CO₂ buildup in my blood leading to hospitalization.”* He now relies on a ventilator mask at night and mouthpiece ventilation during the day.

Sam, whose scoliosis once compressed his heart and lungs, described declining respiratory function despite aggressive supportive care, explaining, *“Today, my lung function is about 22 percent. That's something I think about every day, and it's extremely frightening.”* Angela recalled that a routine cold caused Alyssa to struggle to breathe, ultimately leading to fatal cardiac arrest, emphasizing that for some families the disease ultimately becomes fatal.

Progressive muscle imbalance drives significant orthopedic complications and major surgeries. Sara underwent spinal fusion at age twelve—fusing nearly her entire spine—after which her *“limp became a waddle”* and she could no longer get up from the floor, and both Sam and Justin required corrective spinal surgery for scoliosis severe enough to compromise their lungs.

Hearing, vision, and facial involvement were often the earliest signs of disease. A hearing impairment at age 1 was Justin's first manifestation and disrupted his language development, and he also contends with eyelids that do not fully close; Vera and Angela each described daughters who could no longer smile by age 2, with Vera observing that FSHD *"quite literally steals your smile"* and Angela recounting how her daughter's facial weakness was initially misdiagnosed before FSHD was identified.

Pervasive pain, chronic fatigue, and diminished bone density compound these losses. Sara described relentless fatigue and pain from her body's constant compensation and voiced fear about developing osteoporosis at only age 17, while Justin noted that reduced weight-bearing has left him prone to osteoporosis and fractures alongside pressure sores and tachycardia. Maggie explained that *"my muscles hurt. I push through fatigue and nap often."*

Collectively, participants emphasized that early-onset FSHD is a complex, multisystem disease which requires ongoing multidisciplinary care, and that its most feared complications—particularly respiratory failure—extend well beyond skeletal-muscle weakness.

D. Early-onset FSHD profoundly affects emotional well-being, childhood development, and family life.

Participants described the emotional burden of early-onset FSHD as extending well beyond physical disability. Children and young adults spoke about growing up differently from their peers, describing social isolation associated with progressive disability. As physical function declined, many found themselves unable to participate in sports, recreational activities, school events, and spontaneous outings with friends. Several explained that these exclusions accumulated gradually, resulting in shrinking social circles and increasing feelings of loneliness during adolescence.

Participants described withdrawing from social activities not because they lacked interest, but because inaccessible environments and the need for assistance made participation difficult or emotionally painful. Kate explained that she had begun sitting out activities she loved, including sleep-away camp and house parties, because *"if the house isn't accessible, I end up stuck on a couch alone while everyone else is having fun."* She added, *"It's easier to just stay home than to feel like a project for my friends to manage."*

Maggie similarly described how progressive weakness took away sports and family activities that had once been central to her childhood. She had played softball from a young age and believed she had potential as a pitcher, but explained, *"While my teammates were getting better and stronger, I was getting worse and weaker. FSHD made the sport that I loved since I was 5 unenjoyable and then impossible."* Maggie also described a similar experience with her family, recalling how she recently tried to join a family volleyball game but realized she couldn't participate, *"I sat and watched... Now I miss out on family activities and traditions."* Sara summarized the emotional experience of growing up with FSHD by stating, *"Being a kid and having FSHD, frankly, sucks."* She described persistent fatigue and pain, as well as the fear created by every new moment of weakness. Although she spoke with humor about her experiences, she acknowledged, *"Behind this jading and the jokes, I'm scared."*

Participants also described how FSHD transforms ordinary adolescent milestones into sources of anxiety. Rather than focusing only on academics, careers, relationships, or moving away from home, speakers worried about whether they would be able to dress, bathe, eat, use a bathroom,

navigate a college campus, obtain employment, or live independently. Kate captured this contrast by explaining, *“I’m at the age where I should be excited about college, but honestly? It’s terrifying. I’m not scared of the classes; I’m scared of the bathroom.”* Sam described watching cousins his age attend school, work, build social lives, and become independent while his own opportunities became increasingly constrained. *“It’s not fair that others can strive for what they want while my body makes every dream or passion of mine nearly impossible,”* he said. *“I just want to live just like everybody.”*

Caregivers described their own emotional burden as a form of continual or anticipatory grief. Vera explained, *“We live with constant anticipatory grief—mourning not only what has already been taken, but what may still come.”* Her concerns included whether her daughters would lose additional function, require feeding tubes, or develop life-threatening complications, as well as who would care for them if their parents were no longer able to do so. Angela described the profound uncertainty of caring for her surviving daughter after losing Alyssa to complications of early-onset FSHD. She explained that watching Nessa lose opportunities to attend school, spend time with friends, and participate in ordinary teenage experiences had been *“one of the most heartbreaking parts of this disease.”* After experiencing the death of one child, the possibility of further progression was not theoretical, but a daily and deeply personal fear.

Collectively, participants emphasized that early-onset FSHD reshapes identity, confidence, friendships, education, recreation, and family relationships throughout the lifespan. The emotional burden arises not only from physical impairment, but from repeatedly watching valued activities and anticipated milestones become inaccessible. Patients described wanting the same opportunities as their peers—to play sports, attend social events, pursue education, build careers, and live independently—while caregivers described the grief of knowing that continued progression may place those opportunities increasingly out of reach.

E. Early-onset FSHD imposes intensive, lifelong caregiving demands and substantial educational and financial burdens on entire families.

Participants made clear that early-onset FSHD imposes intensive, lifelong caregiving demands and substantial educational and financial burdens that extend well beyond the individual patient. As function declines, patients require growing assistance with the most basic activities of daily living, while families reorganize their lives, careers, and finances around that care.

Justin described requiring aides for most daily living tasks and the care of seven specialist physicians, along with prescriptions and frequent wheelchair repairs, concluding that the cumulative costs *“add up and are unaffordable.”* Although he earned a PhD and worked as a research scientist studying FSHD, he explained that the accommodations he needed to keep working were *“not sustainable in the long run.”* Sam recounted submitting more than forty job applications and being eliminated from the two interviews he obtained because he could not meet the physical requirements; he also described needing his mother to get him out of bed, dress, bathe, and take him to the toilet.

Caregiving parents described around-the-clock responsibility. Vera’s daughters depend on the family for nearly every aspect of daily life, including eating, bathing, and dressing, and every school event requires weeks of advance planning. Angela provided total care for Alyssa, who was

never able to care for herself, and now assists Nessa, who has had to move from in-person to online school.

The disease also reshapes education and early careers. Sara, a high school senior, described her fear of entering the workforce, questioning whether she can *“even add value to the team with all the exceptions and modifications... that I need to function,”* while Justin’s schooling was repeatedly disrupted by speech therapy, inaccessible buildings, and mobility limitations.

Collectively, participants conveyed that the burden of early-onset FSHD is borne by the entire family—through lost income and employment, the financial strain of specialized care and equipment, disrupted education, and the physical and emotional toll of round-the-clock caregiving. These cascading caregiving, educational, and financial consequences underscore that the value of a disease-modifying therapy extends well beyond the individual patient.

F. Existing management is only supportive; there is no therapy that halts disease progression.

Every panelist emphasized the fundamental inadequacy of the current treatment landscape. There is no FDA-approved therapy that addresses the underlying cause of FSHD, slows muscle loss, or halts disease progression. Patients and families are left with vitamins, physical therapy, adaptive equipment, and surgical interventions that manage symptoms but do nothing to alter the disease trajectory. This therapeutic void is particularly devastating for families of children with early-onset FSHD, who watch their children lose function with no roadmap and no meaningful intervention available. The absence of treatment options stands in stark contrast to the well-characterized molecular mechanism and the clear therapeutic target it represents.

Panelists uniformly described the available options as inadequate. Vera recounted being told there was *“nothing we can do,”* with *“no meaningful treatment options. No roadmap. No relief. Only progression.”* Sara explained that, having never been in a clinical trial, *“all I have to slow this down are like vitamins and some at home exercises... They don’t stop muscle death, and they don’t reverse muscle loss.”*

Collectively, participants described a treatment landscape that tries to manage symptoms but does nothing to alter the course of the disease, leaving patients to watch function disappear despite a well-characterized molecular target. They emphasized that the current therapeutic void reflects not a scientific dead end, but an urgent unmet medical need required accelerated development and access for a population that has no time to wait.

G. Patients and families are highly motivated to participate in clinical research despite uncertainty and risk.

Participants universally expressed strong interest in participating in clinical trials investigating disease-modifying therapies. They viewed clinical research not only as an opportunity to potentially benefit themselves but also as a responsibility to future generations of patients.

Participants consistently indicated they would accept substantial inconvenience, uncertainty, travel, and reasonable treatment-related risks because the alternative is continual disease progression without effective therapy. This willingness reflects an acceptance of risk that applies

not only to investigational therapies but also to approved treatments that have not yet been studied in pediatric or early-onset populations.

All seven panelists expressed strong, universal interest in participating in a future clinical trial for FSHD. Their enthusiasm was unequivocal, reflecting both the severity of their unmet need and their acceptance that investigational therapies carry inherent uncertainty. At the same time, panelists articulated thoughtful, specific considerations that would inform their enrollment decisions.

Several panelists raised concerns about whether participation in a gene therapy trial could jeopardize eligibility for future treatments. Vera explained that her daughters would participate in any opportunity available but that her primary apprehension concerned whether a gene therapy (as opposed to, for example, an antisense oligonucleotide that clears the body) could foreclose access to future options. She emphasized that she would carefully weigh the available data but that it was not an absolute barrier to participation. Angela expressed a similar concern, noting that trials fill up quickly and she would make participation happen for Nessa regardless of travel, but that she would need to understand both the expected benefit and whether the therapy might preclude access to future treatments. Justin expressed a similar concern, specifically around AAV exposure that may make him resistant to future AAV therapy.

Safety and understanding of expected benefit were also central concerns. Kate stated she would be very excited to participate but wanted robust safety data, particularly for gene therapy, and needed to understand how a therapy would actually help and whether it could worsen her current quality of life; she expressed a preference for later-stage trials with more accumulated data. Sam emphasized that before committing significant time, effort, and travel, he would want to understand the likely magnitude of benefit and expressed concern about being assigned to placebo for the duration of a trial.

Practical considerations also factored into panelists' decisions, especially considering the school-age of several participants. Sara noted that as a high school senior, she would want to consider location and disruption to her final year, though she would travel out of state if required. Maggie confirmed she had already attempted to enroll in a trial and would try again; she was aware of opportunities outside the United States and would travel, though timing was difficult given her senior year and college plans. Justin indicated that he would be very willing to participate but he would want to know about the safety profile, including the risk of a serious adverse effect, of any potential drug.

Participants consistently framed trial participation as both a personal opportunity and a contribution to the broader FSHD community. Several stated that although they hoped to benefit individually, they were equally motivated by the possibility of improving the lives of younger children and future generations diagnosed with FSHD. The universal willingness to participate, even in the face of significant apprehension about safety and foreclosed future access, underscores that patients and families with early-onset FSHD accept substantial risk because the alternative—continued disease progression without treatment—is worse.

H. Current clinical trial eligibility criteria leave many patients with the greatest unmet medical need without access to investigational therapies.

During both the panel presentations and moderated discussion, participants described significant barriers to clinical trial participation despite closely following therapeutic development.

Participants expressed frustration that individuals with early-onset disease often are ineligible precisely because their disease progresses before trial opportunities become available. As Sam summarized, *“We’re excluded when we’re younger because of age. Then when we’re older because of disease severity. We’re caught in a system that never gives us a chance,”* highlighting that treatments must reach patients before significant muscle loss and irreversible disability occur. Pediatric trials are therefore essential. Preserving muscle from a young age could delay progression, maintain independence, and ultimately prevent the severe disability that later becomes a reason for trial exclusion.

Maggie’s experience illustrated the dilemma of very narrow inclusion criteria. She postponed major milestones during her junior year of high school because participating in a clinical trial represented her greatest hope of preserving muscle function. After completing extensive screening, she was ultimately told she did not qualify because she *“moved too fast”* during functional testing. Reflecting on that experience, she said, *“My three brothers can walk faster than me, including the one nine years younger. I need an ankle brace and sometimes fall while walking. My arms barely move. My muscles hurt. I push through fatigue and nap often. And somehow... I was not affected enough.”*

Others noted that current eligibility criteria exclude pediatric patients, non-ambulatory individuals, and those with advanced disease despite their substantial unmet medical need.

Participants emphasized that the greatest obstacle is not awareness of clinical trials but the inability to qualify for them. Several expressed hope that future studies would include individuals across the full spectrum of FSHD so that children and severely affected patients are not left behind.

I. Meaningful treatments do not need to restore lost function to have transformative impact.

Participants consistently stated that a meaningful therapy does not need to reverse the progression of FSHD to dramatically improve their lives. Across ages and disease stages, preserving existing function, slowing progression, and maintaining independence were identified as the most important treatment goals.

Maggie explained simply, *“I hope to receive a treatment soon so I can keep the function I still have.”* Sam emphasized that despite being non-ambulatory, he still has important muscle function worth preserving, stating, *“I still have a lot of muscle to lose... I at least want to hold on to the function I have now—or maybe even regain some of what’s been lost.”*

Kate similarly highlighted the urgency of therapeutic development, explaining, *“I don’t have time to wait for a perfect future—I need a world, and treatments, that can keep up with me today.”*

Collectively, participants emphasized that even modest slowing of disease progression would meaningfully preserve independence, education, employment, respiratory function, and quality of life.

Panelists were asked what they would most want a future therapy to achieve, even if it could not fully cure FSHD. Responses consistently reflected that slowing or stopping disease progression would be profoundly meaningful, even without restoring lost function.

Angela expressed that slowing or stopping progression, particularly of mobility in both upper and lower limbs, would be her primary goal. She contrasted her daughter Alyssa, who required assistance with transferring and bathing before she passed away, with her daughter Nessa, who does not yet need that level of help and can still drive—illustrating the concrete difference that preserved function makes in daily life.

Justin stated that stopping progression would allow him to maintain his current level of independence, and would likely prevent worsening of other symptoms such as hearing, vision, and breathing. He added that, ideally, he would want a therapy that restores some muscle mass and function.

Sara described wanting to slow progression to preserve her relative independence—her ability to wash, dress, and feed herself—and ideally to live independently in college. Maggie noted that slowing or stopping progression would be meaningful because while she has adapted to her current symptoms, she knows they will worsen, and she wants to preserve remaining muscle function, particularly in her left ankle since she has already lost function in her right.

Vera emphasized slowing or ideally stopping progression and addressing the chronic fatigue that causes young girls with FSHD to miss school from exhaustion. She invoked the phrase “time is muscle” to convey the urgency of intervention. Kate stated that slowing progression would be a minimum acceptable outcome and that stopping progression at her current level—even without regaining full independence—would be acceptable, though she expressed hope for some muscle function restoration.

Sam focused on stopping progression and building respiratory muscle capacity, particularly through his diaphragm and the ability to cough, to prevent declining lung capacity and to help clear mucus during illness—reflecting the life-threatening respiratory dimension of his disease.

Taken together, these responses demonstrate that patients and families do not functional gains as the threshold for meaningful benefit—simply stopping progression and preserving what remains would be transformative. The earlier in life, the better.

J. Families face challenges in accessing clinical research, along with systemic barriers to trial enrollment

During the FDA question-and-answer portion, Agency staff acknowledged their awareness of the challenges in participating in clinical research and asked how frequently panelists had discussed clinical trials with their treating physicians. Angela responded that, despite both of her daughters having been diagnosed in 2011, the first time a doctor had raised the topic of a clinical trial with her was in the past three years since moving to Tennessee. Sara noted that even in California—home to multiple universities and clinics involved in FSHD research—she has not been able to

access any clinical trials, including during the past two years in which she has been age-eligible (since turning 16). Vera described how getting a diagnosis alone was an ordeal and that no clinician has ever discussed clinical trials with her family; she has had to travel six hours to Rochester for Isla and Siena to receive specialized care.

Agency staff also asked how panelists find out about clinical trials and what would make it easier for patients to learn about available research opportunities.

Vera explained that she is aware of clinicaltrials.gov and does not believe the fundamental problem is awareness—patient advocacy groups are effective at publicizing available trials and emerging data. Rather, the problem is trial eligibility criteria, which are so narrow that even patients who are aware of trials and eager to participate cannot qualify. Ally Roets, a parent and patient advocate from the FSHD Cure Initiative, concurred, noting that the challenge is not awareness but that inclusion criteria remain so restrictive that even enrolling an age-eligible 16-year-old in a trial has presented significant difficulty.

Taken together, participants described a clinical research landscape in which awareness of trials is no longer the primary obstacle. Instead, they emphasized that delayed referrals, limited physician engagement, and restrictive eligibility criteria leave many patients with early-onset FSHD unable to access clinical research despite being informed, motivated, and eager to participate.

VIII. Patient/Caregiver Testimonials

Sara, 17 years old, living with FSHD

- **San Diego, CA**
- **High school student**
- **Diagnosed with FSHD at 8 years old**
- **Spinal fusion**
- **Can't run, jump, or stand longer than 30 minutes**
- **Limited facial expression and function**
- **Limited arm function**
- **No clinical trial experience**



Hello everyone, it's good to be here. My name is Sara, I'm from California, and about two months ago, I turned seventeen. Believe it or not, I have *always* wanted to be seventeen. In every high school hijinks comedy, the main characters are all seventeen, you've got 'Dancing Queen' by ABBA, 'Edge of Seventeen' by Stevie Nicks. Driver's licenses, house parties, biking to the mall, sprinting into the ocean with youthful abandon. I mean, hasn't the world always kind of been made for being seventeen?

Well, for some people, absolutely, but for me and my community of young people with Early Onset FSHD, no, not really.

When I first got my diagnosis as eight years old, [seven maybe? CHECK] I thought my life was over. And could you blame me? The social worker delivered the news like a death sentence, in hushed tones and sad eyes. I'd say my tearful response was appropriate for someone who was barely learning how to multiply. But, I'm getting a little ahead of myself.

I was probably four years old and less than four feet tall when my mom noticed that my smile didn't look like most other smiles. It was small, barely a glint of front teeth to show my joy. I remember on picture day, we'd all funnel into the auditorium class by class, wait in line with our ribbony dresses and collared shirts. Flash by flash the line in front of me would get shorter, and as I sat with my tiny shoes in the precise angle of the footprints stickered to the floor, I'd smile my best smile.

The cameraman would tell me to smile *more* so I did. I stretched my face until it hurt, and even then, that wasn't enough for him. He'd sigh, snap the shot, and move me along. I would leave every picture day with one more chip in my childlike confidence—you know, the one you never question when you're small, but grow up and wish you'd never let life kick it out of you?

Then, it was chewing with my mouth open. I was a little older, and we were at an Easter brunch. I was minding my business and my plate, when my dear uncle told me to chew with my mouth closed, or else I'd catch flies in it. Looking back now, it was funny, but to seven-year-old me, who was already the non-consenting monkey in the middle, already last in the laps around the blacktop,

already being told to smile wider, write faster, kick harder; it'd only broken me, if not just a little more.

So when I was twelve, with a curve in my spine and a limp in my walk, and I was informed of a treatment in the works— a cure— and it seemed like, if anything, what would save me. But a viable treatment was still a few years away, and the burning spasms snaking through my back were only getting worse, so we settled on spinal fusion surgery as salvation instead. My whole spine, actually, with only three vertebrae left unfused. My limp became a waddle and my ability to get up from the floor? Gone.

For me and others with FSHD, that happens after only two weeks in a hospital bed.

I don't want to sound jaded, it's just that being a kid and having FSHD, frankly, sucks. And I could pivot, and talk about all the ways I am so blessed to have the support of the FSHD community, of my family, and of my friends, and the unique perspective my body given me, but, I'm tired. No, I'm literally tired, because I get fatigued easily, because my muscles *die*. I'm in pain, because all parts of me are compensating for each other, right now, because my muscles *die*. And every moment of weakness makes me fear for my future. Is my split-second of bad coordination with getting a spoon to my mouth a fluke? Or will I, in three years be unable to feed myself?

Because, behind this jading and the jokes, I'm scared. I've never been in a clinical trial, all I have to slow this down are like vitamins and some at home exercises. They can address pain, stiffness, and not exactly slow down muscle death, but make each period of it leave less of a dent. Two capsules from three bottles, once a day. Two sets of ten reps, three times a day, three times a week. Forever.

Don't get me wrong I love taking care of my health, and moving around, but to do it from a place of fear can't be healthy. These treatments don't address waning bone density, they don't stop muscle death, and they don't reverse muscle loss. They don't guarantee me a safe walk down a hill, a rail-less climb up the stairs, or grant me a spontaneous swim in the ocean with my friends.

Not only that, but I don't know how I'm going to be independent as a young adult. I can cook for myself well enough, I can dress and bathe myself, if I really wanted to I could clean around the house, just, don't tell my mom. But say I'm strolling down the street at night in my electric scooter; like most girls, I carry pepper spray, but unlike most girls, I'm 90 pounds and my punches aren't the most effective. That scares me.

Osteoporosis at 17? That scares me. Getting a [gasp] job? That scares me, and not just in the normal teenage way, in the way that makes me question if I can even add value to the team with all the exceptions and modifications and commas that I *need* to function.

Because I like poetry, I can tell you that some days I feel like a living corpse, tired, beaten, and with dead muscle turned fibrous fatty tissue in my arms and legs. But because I can't always disguise everything with pretty words and alliteration, I'll just wrap up with:

It's really good to be here, my name is Sara, I'm from SoCal, I can stand for 30 minutes before I need to sit down, I can't jump, I can't reach my arms above my chin, I love the ocean, I love horseback riding even though it's challenging, I can't really pick things up, I have not run in a decade, and I want my seventeenth year on this earth to be a good one—I think it will be. I'm just counting on the FDA to make my eighteenth year great, and hopefully, to make another small-smiled, slow running, eight-year-old girl's *life*, a great one too.

Sam, 20 years old, living with FSHD

- **Tucson, AZ**
- **College student/audio engineer**
- **Diagnosed at 6 years old**
- **Multiple surgeries**
- **Lost ambulation at 13 years old**
- **Restrictive lung disease**
- **No clinical trial opportunity**



Good morning, and thank you for the opportunity to speak today.

My name is Sam. I'm 20 years old, and I live in Tucson, Arizona. I was diagnosed with FSHD when I was six years old, but the signs were there from the beginning. I was born with hearing loss, had speech delays, and my body didn't move like other kids.

As I grew up, my symptoms progressed quickly. By age eight, I had foot drop in both feet and needed braces to walk. I went through two tendon transfer surgeries to hold on to my mobility, but my muscles kept getting weaker. By eleven, I needed a walker, but I still got tired walking between classes and kept falling, so I started using a wheelchair for longer distances. By thirteen, I was using a wheelchair full time and I could no longer stand or walk a few steps.

At fifteen, I had a complete spinal fusion because scoliosis was compressing my heart and lungs. The recovery was difficult, and I lost even more physical function. It took my ability to transfer, raise my arms higher, and continues to cause some back pain. Since then, my breathing has continued to decline. I've had pneumonia several times. Even a common cold can be dangerous. I have learned to use a nebulizer and cough machines early to try and prevent mucus from settling in my lungs. Today, my lung function is about 22 percent. That's something I think about every day, and it's extremely frightening. I do lung expansion therapy and wear a bi-pap at night to hopefully prevent that number from getting lower.

When I was in eighth grade, I won a national writing contest called Breaking Barriers, sponsored by the family of Jackie Robinson. I remember in my essay, I wrote about a new treatment for FSHD that was in clinical trials. I was excited. I naively believed that I would have a chance to participate—that maybe I could slow down the disease for myself and contribute to finding a cure.

But I was told I couldn't join because I was under 18. I remember thinking to myself, "That's okay—I'll wait."

But by 18, I had lost the ability to walk, and I learned that I still didn't qualify—because I was no longer ambulatory.

Now I'm being told that even if a therapy were to be approved, it may still be difficult for someone like me to access it, because there won't be enough safety or efficacy data in people who are more severely affected.

So where does that leave me and patients like me?

We're excluded when we're younger because of age. Then when we're older because of disease severity.

I still have a lot of muscle to lose, and I don't understand why the system seems to assume that people in wheelchairs don't. I at least want to hold on to the function I have now—or maybe even regain some of what's been lost.

This is not just frustrating—it's devastating.

I go to physical therapy to maintain what strength I have, but it can only do so much.

Today, I attend community college and live with my mom and stepdad. I have clear goals for my future—I want to work as an audio engineer. I already volunteer for local venues, and I want to turn that passion into a career. There's an audio school in Phoenix that I hope to attend. After touring the school, I found that there are some things I can and cannot do in this field, like lifting heavy speakers, but there are plenty of tasks I can do from behind a sound board or on a laptop. If I lose even more function, these easier tasks will become just as impossible, and I will have to give up on what I've been wanting to do since 8th grade.

Even now, I can't find a part-time job. I've put in over 40 job applications. I got two job interviews and I was eliminated from both because I couldn't meet the physical requirements. One was an audio position I was qualified for and really wanted, but I had to be able to move speakers. It crushed me to tell him that I couldn't do it. I left the interview defeated and angry.

I worry about how much strength I will lose, and how I'll be able to provide for myself. I don't want to be a burden on the government or my family. I already need help with so many things that affect my independence. Being able to cook, clean, and care for myself is something I fear I will never be able to do independently. I'm a 20 year old adult who needs his mom to get him out of bed, dress, bathe, and take him to the toilet.

I look at my cousins, who are my age. They're in school, working part-time jobs, building social lives, and becoming independent. It's not fair that others can strive for what they want while my body makes every dream or passion of mine nearly impossible. I just want to live just like everybody.

Maggie, 17 years old, living with FSHD

- **Ann Arbor, MI**
- **High school student**
- **Diagnosed at 13 years old**
- **Foot drop**
- **Limited arm function**
- **Forced to quit hobbies and sports due to disability**



My name is Maggie, I am 17 years old and I was diagnosed with FSHD at 13. FSHD affects every area of my life: at school, at home, in friendships, and with hobbies. It affects me in significant ways physically, but also emotionally and socially.

I now know I was lucky to have experienced a fairly typical childhood when I was younger. I was very active and independent. And then the rug was pulled out from under me. And then again, and again. And it will keep happening until I have access to a treatment.

I have had facial weakness since I was 4, which I later learned from pictures. By 10, I had trouble with my posture and couldn't figure out why. At 13, I began to lose strength in my right arm. Suddenly it became very hard to wash hair, reach shelves and put on shirts.

Since my diagnosis, I have lost the ability to raise either arm, walk normally, or run at all. My loss happened in stages, with a huge new loss striking me every year: my right shoulder, my core, my right ankle, and then my left shoulder.

Imagine moving around throughout the day with your arms tied to your waist so you can't raise them above your chest. It's very hard to function. However, I've become a problem-solver so I can continue to do as much as possible. I walk my fingers up walls or use momentum to fling my arms so I can try to use them.

At this point, it is difficult and tiring to even lift food or drinks to my mouth, and it's hard to carry books or raise my hand at school. I can't control my right ankle, and my legs get tired after walking even short distances. I am always tired. These difficulties remind me daily that I have a progressive disease.

I started playing T-ball in kindergarten, and I played baseball and then softball. I had good potential as a pitcher. But eventually, while my teammates were getting better and stronger, I was getting worse and weaker. FSHD made the sport that I loved since I was 5 unenjoyable and then impossible.

There is so much loss with FSHD. I used to water ski with my family, and I would get up easily. Two years ago, I tried to get up on water skis a dozen times (with my mom and grandma trying to

physically help me) before letting that activity go. It was devastating and embarrassing to fail again and again while my little brother succeeded so quickly.

Recently, I was excited to play volleyball with my cousins at a family party. I was nervous that I wouldn't be able to but gave it a try. I quickly realized that I wouldn't be able to participate and I'd just embarrass myself. I sat and watched. Now I miss out on family activities and traditions.

Pieces of me have slipped away and the activities I'm grateful to hold onto are next.

I now have far from a typical childhood. And I won't experience a typical adulthood. I have fewer career opportunities and face barriers and expenses because of FSHD.

FSHD is also socially isolating. I miss out on spending time with friends, and I'm reminded of the time they share together when they have inside jokes or talk about their experiences that I can't take part in. It's hard enough to lose abilities, but it's worse when I know it affects my friendships too.

FSHD also causes anxiety because I never know when another muscle will suddenly start rapidly deteriorating. The loss honestly feels like it happens overnight. It's also really stressful to be unable to function like other classmates. One of my biggest fears is to be stuck in an active shooter situation. I can't defend myself, and I'm sure that I'm the absolute slowest person in the entire school.

I had a period of great hope. I qualified for a clinical trial screen and took the opportunity and obligation seriously. I put literally everything in my life on hold for the one thing that might help me. The important activities of junior year, like the ACT, college applications, and AP courses, took a back seat to trying to get into a trial to save my muscles.

I flew to my screening site with excitement (and fatigue that follows me like a shadow). I knew that I had a 50% chance of going through all the challenges of a clinical trial and then being given a placebo. But it was my only option. I was at the screening appointments for hours, and after I completed testing, I was told that I moved too fast to qualify. I failed the screen.

My best friends are all on the school cross country and track teams. My three brothers can walk faster than me, including the one nine years younger. I need an ankle brace and sometimes fall while walking. My arms barely move. My muscles hurt. I push through fatigue and nap often.. And somehow... I was not affected enough. I sobbed after hearing that news. My parents and I felt depressed and hopeless for months.

I hope to receive a treatment soon so I can keep the function I still have. And I hope that it'll come soon for young kids so that my 10-year-old brother, Peter, won't experience the physical and emotional pain that I've had to endure.

Thank you for listening to my story.

Vera, mother to Isla (10 years old) and Siena (8 years old)

- **Bayonne, New Jersey**
- **Elementary students**
- **Diagnosed at 5 and 3 years old**
- **Speech difficulties**
- **Lost ambulation at 9 and 8 years old**
- **Upper limb weakness**



My name is Vera, and I live in Bayonne, New Jersey, with my husband and our two daughters, Isla, who is 10, and Siena, who is 8.

In October 2021, when my daughters were just 3 and 5 years old, our world changed forever. That was the first time I heard the words Facioscapulohumeral Muscular Dystrophy—FSHD.

But the truth is, FSHD had already been part of my story long before that. I just didn't know it.

I was diagnosed with FSHD at 35 years old. Looking back, the signs had always been there: hearing loss, an asymmetrical smile, and never being able to run for long periods of time. They were things I adapted to and brushed aside—things no one ever connected together.

Until my daughters.

Isla and Siena were born healthy, joyful, fearless, and full of life. Isla loved dance and tumbling. Siena followed closely behind her big sister with endless energy and curiosity.

At first, the signs were subtle.

Hearing loss.

Then facial weakness.

And by the age of two, both of my daughters could no longer smile.

We spent years going from specialist to specialist—MRIs, genetic testing, and countless appointments—desperately searching for answers.

Eventually, we got one.

From that moment on, our lives split into before and after.

What makes early-onset FSHD so devastating is not only how severe it is—it is how quickly everything can disappear.

Just two years ago, at ages 5 and 7, my daughters were still dancing.

When dance season began that September, Isla was keeping up with every other child in the studio. She was running, jumping, dancing through rehearsals, and doing everything her peers were doing.

But over the next ten months, we watched FSHD take more from her.

As rehearsals continued, standing became harder. Her balance became increasingly unsteady. Week by week, we watched her fight to do the things that had once come so naturally.

By recital day in June 2024, at just 8 years old, bars had to be placed around her on stage to keep her from falling during a routine that lasted only a few minutes.

As a mother, there is nothing that prepares you for watching your child fight just to stay standing while trying to hold onto the childhood she loves.

Today, Isla can no longer walk.

In just 24 months, she lost all mobility.

Then it happened to Siena.

This past March, Siena was still walking. One morning, at just 8 years old, she told us she didn't want to go to school. I immediately knew why.

She was terrified of falling.

That day was March 23, 2026.

We didn't know that morning would be the last day our daughter would ever walk.

If we had, we would have held her a little longer before she got out of the car.

At just 8 years old, Siena has not walked since.

One day she walked into school.

The next, she never walked again.

Today, neither of my daughters can stand independently or walk. They depend on us for nearly every aspect of daily life.

They need help eating. They have never been able to shower independently or do their own hair. They cannot simply walk into their rooms, choose their own clothes, or spontaneously join activities with friends like other children their age.

Every school event requires weeks of planning, and despite our best efforts, they have still missed countless field trips because they were inaccessible.

FSHD is cruel.

It does not just rob children of physical strength—it steals independence, dignity, and childhood itself.

It is a disease that quite literally steals your smile.

On the hardest days, the grief feels suffocating. You never truly learn how to cope because this disease never stops progressing. There is always something more to lose.

One of the most devastating parts of this journey is being told there is nothing we can do.

We are sent home and told to come back every six months—not for treatment or solutions, but simply to document how much worse things have become.

There are no meaningful treatment options.

No roadmap.

No relief.

Only progression.

Sometimes it even feels like the few interventions we are offered, such as physical therapy, leave them even more exhausted.

We are simply watching this disease take more from our children in real time.

We live with constant anticipatory grief—mourning not only what has already been taken, but what may still come.

Will they lose their vision?

Will they one day need feeding tubes?

And beneath all of those fears is the question that never leaves us:

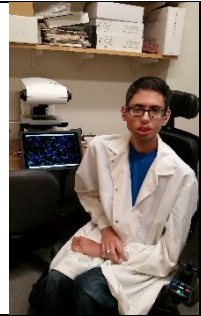
Who will care for them if something happens to us?

The heartbreaking reality is that, because of how aggressive their disease is, we also live with another unimaginable fear—that we may have to bury our daughters.

No parent should ever have to carry that thought.

Justin, 35 years old, living with FSHD

- **New Haven, CT**
- **Molecular and Cell Biology & Genetics PhD**
- **Diagnosed at 4 years old**
- **Speech difficulties**
- **Requires aide to assist with daily living activities**
- **Unable to continue career due to severe disability**



My name is Justin, I am 35 years old and live in New Haven, Connecticut. I was diagnosed with FSHD at only 4 years old. My first symptoms were evident even at the age of 1, manifested as a hearing impairment. I would hear my dad's deep voice but ignore my mom's higher pitch. Accordingly, my language development was impaired, requiring lots of disruption to my education as symptoms developed. For example, my muscle weakness caused a speech impediment, resulting in me needing to take time away from my classes to work with a speech therapist in the middle of the day several times a week from elementary to high school. Further disruption occurred once my ambulation became affected. I started to develop a limp at 10 years old, which quickly led to crutches by 11. Wheelchair dependence occurred by 13. Consequently, I needed to leave each class a few minutes early in order to reach the next one on time, making me miss the end of each lesson.

As my symptoms continued to worsen, it influenced my higher education as well. It was very important I attend a college relatively close to home so that my parents were available if needed. While I was lucky to live in the northeast with lots of options, ultimately attending Tufts University, there were still significant challenges. First, northeast winter brought lots of snow. This made getting to class difficult and there were cases when the weather caused me to miss class entirely. Furthermore, most northeast colleges are very old and mine was no exception. Most buildings were not handicapped accessible and classes would have to be relocated to unideal locations for me to attend. This affected me socially as well since many student clubs were inaccessible nor were the dorms and houses my friends resided in.

There have been numerous other ways in which this disease affects my health. Most people are familiar with the loss of ambulation those with early onset FSHD typically experience, leading to becoming wheelchair bound. However, the severity of muscle wasting that leads to a wheelchair results in many other secondary symptoms. The loss of weight bearing reduces bone strength, making me prone to osteoporosis and increased risk of fractures if I fall. My legs have become contracted so they don't stretch and blood flow to my feet is poor. Pressure sores form from sitting all day. I have tachycardia from the reduced activity muscle weakness necessitates. My eyelids don't close all the way, requiring me to use ointment at bedtime. However I still sometimes get irritated eyes and scratches on my cornea. I had scoliosis so bad it affected my lung capacity and I needed spinal surgery to correct it. Most concerning was an incident last summer where I almost

died. Breathing issues caused a large CO₂ buildup in my blood leading to hospitalization. I now use a ventilator mask every night and mouthpiece ventilation during the day.

As you can imagine, the costs to deal with these symptoms both monetary and through time commitment are significant. This includes 7 specialist doctors, prescriptions and frequent wheelchair repairs. Most significantly, I require aides to assist me in most daily living tasks. Altogether, these costs add up and are unaffordable.

In order to offset these costs, I try to work. I was actually able to achieve a PhD while my symptoms were more manageable and worked as a scientist studying FSHD at Yale. However, to do so required many accommodations that were not sustainable in the long run. With the severity of my symptoms I do not think it is possible for me to work, even with my education level. Voice to text tools to overcome typing issues don't work because of my speech impediment. Careers that focus on talking to people directly instead become an issue due to my hearing impairment and breathing issues that affect my speaking endurance. As my FSHD continues to progress I worry about what my future looks like. A greater loss of my independence could result in assistance being required for every single movement and function that most people take for granted. A treatment for FSHD is thus critical.

Kate, 15 years old, living with FSHD

- **Boston, MA**
- **High school student**
- **Diagnosed at 6 years old**
- **Lost ambulation at 9 years old**
- **Limited arm function**
- **Spinal deformity and contractures**



My name is Kate, I'm 15 years old from Massachusetts and I was diagnosed with FSHD at six years old. My first memory is skiing at 18 months old. People usually find that impressive, but for me, it's a bit bittersweet. Because FSHD started so early for me, I didn't get a "normal" childhood and then a diagnosis. I was having muscle issues before I even knew what a "muscle" was. While other kids were learning to tie their shoes, I was subconsciously learning how to handle loss.

Growing up with FSHD means I don't really look back and mourn a "healthy" version of myself. I never knew her. Instead, I live in a constant state of looking forward—but not in a hopeful "daydreaming" way. I look forward and wonder, "What is the next thing I'm going to lose?" Because with FSHD, something is always being taken away.

My childhood was measured not by milestones of growth, but by a timeline of losses. At three years old, I lost half my smile.

Throughout my early childhood, I could never run as fast or keep up with the other kids. I fell down constantly, and "I'm okay" became my automatic catchphrase before anyone could even ask.

By age eight, the progression accelerated. I slowly watched my ability to raise my hands above my head slip away.

And at age nine, I stopped walking independently altogether.

Since then, the progression has taken aim at my core, weakening my abs the most. But it's the small, quiet losses that happen month-to-month that remind me I am always on the clock. Just a few months ago, I looked down and realized I could no longer lift my thumb to give a full thumbs-up. In school I would sometimes get scolded for rolling my eyes at a teacher when in reality I couldn't close my eyes all the way when I blink.

I live in a world that wasn't built for me, so I've had to become a professional problem-solver. I've spent my life finding "stealth" workarounds. When I was losing strength in my legs, I would find a way to lean against a table so it would look like I'm just being casual instead of trying not to fall over. I've become an expert at hiding the struggle because I don't want to be "the disabled girl"—I just want to be a girl.

But the workarounds are getting harder. My brain has become a GPS for accessibility. Before I go anywhere, I have to run a mental checklist: Can I get into the house? Is there a bathroom I can actually use? Am I going to have to ask my friends to lift me? When you're 15, you want to be independent. But FSHD forces me to be a burden, or at least feel like one. I've started sitting out on things I love—like sleep-away camp or house parties—because if the house isn't accessible, I end up stuck on a couch alone while everyone else is having fun. It's easier to just stay home than to feel like a project for my friends to manage.

There's a weird disconnect in my life. Mentally, I'm probably more independent than most adults. I've had to be. I've had to navigate medical conversations, physical limitations, and social isolation since I was a little kid. But physically, I'm dependent.

I have an independent mindset in a dependent body.

I'm at the age where I should be excited about college, but honestly? It's terrifying. I'm not scared of the classes; I'm scared of the bathroom. I'm scared of how I'll navigate a campus when I'm already struggling to navigate my own life. Even the way I dress is a reminder that I'm not in control. I wear loose, baggy clothes—not because that's my "vibe," but because they're the only thing I can manage by myself in a bathroom stall. I hate tight shirts because they show how much muscle I've lost, making me look "too skinny" in a way that feels fragile. I'm a teenager; I want to care about fashion, but instead, I have to care about ergonomics.

FSHD also changes how people see me. I've noticed there are basically three types of people:

1. People who are scared of me (or just don't know where to look).
2. People who treat me like a baby or a small dog—lots of head-pats and "you're so brave" in a high-pitched voice.
3. People who can look past the wheelchair and see me for who I am.

I love my friends who accept me, but as I get older and move into new chapters of life, I worry about how many of them I'll actually find. When I was little, I was a total extrovert. I did Irish dance and gymnastics. But when I couldn't do those anymore, I lost that connection to my peers. I've become an introvert by necessity, and I've gotten pretty used to being alone[SW3] .

While the medical and drug approval world moves at its own pace, my muscles are on a clock. To manage my current symptoms I have my full time electric chair – can't go anywhere without. I also go to OT and have a personal trainer a few times a week, which is helpful but I don't get nearly the growth others do from working out. We monitor my lungs, heart and muscles on a 6 months basis which means a lot of testing, doctors, and blood work. I don't have time to wait for a perfect future—I need a world, and treatments, that can keep up with me today.

Angela, mother to Alyssa (passed away at 16 years old) and Nessa (17 years old)

- **Nashville, TN**
- **High school student**
- **Diagnosed at 6 and 2 years old**
- **Upper and lower limb weakness**
- **Hearing loss**
- **Respiratory/diaphragmatic weakness**



My name is Angela [] and I'm the mother of two daughters with early-onset FSHD. My daughter Alyssa was diagnosed at age 6, and my younger daughter, Nessa, was diagnosed at age 2. We're originally from New York and now live in Nashville, Tennessee.

When Alyssa was born, she failed the newborn hearing screening. Because my parents are deaf, I was offered genetic testing at the time, but I declined since I was already familiar with hereditary hearing loss in my family. Even if I had pursued testing, it would not have identified FSHD, as it is not part of standard genetic screening unless specifically tested for.

In her first year, most things seemed typical except she did not smile. It was noticed, but we were told she was a late bloomer. By age three, she was still unable to smile. Over time, we started noticing other things. She tripped frequently and struggled to keep up with other children during physical activities. She was initially diagnosed with facial paralysis and possible Moebius syndrome, which led to a facial grafting to restore the ability to smile. This was done in two separate twelve hours surgeries, one when she was 5 and one when she was 6. In her second surgery, she faced complications due to the anesthesia that resulted in a life-threatening event, and she was ultimately diagnosed with FSHD, which also led to Nessa's diagnosis.

Nessa's symptoms were less obvious early on and developed more gradually at first. But as she entered elementary school, the progression became more noticeable - her facial weakness became more prominent, and her gait started to change. Now she is a senior in high school, and her scoliosis and lordosis have progressed to the point where she can only walk very short distances, less than 10 feet, and even then with great care.

She is still able to care for herself in many ways, but those abilities are becoming more limited over time. Because of her mobility, she is no longer able to attend school in person and is now enrolled online. Socially, she wants to be included like any teenager, but her physical limitations make it harder for her to participate in the same way as her peers. Every outing requires someone to help her transfer and get around. As her mother, I gladly do it, but I also know it's a responsibility that many teenagers are not prepared for. Her friends care about her and want to include her, yet the reality of her physical needs can make getting together more difficult. Watching those opportunities quietly disappear has been one of the most heartbreaking parts of this disease.

Alyssa's condition, in contrast, was severe and progressed quickly. By third grade, she was in a wheelchair. She had significant facial weakness that affected her speech, and over time she developed severe weight loss that required a feeding tube. She was never able to care for herself, and I was responsible for all aspects of her daily care.

As she grew into her teenage years, her world became increasingly limited by the disease. Friendships were harder to maintain, fatigue was constant, and many typical teenage experiences were out of reach. Both of my daughters have experienced severe forms of FSHD, but in very different ways, with different timelines and different losses of function.

On September 28, 2021, Alyssa got sick and was struggling to breathe. She went into cardiac arrest and collapsed in my arms. One week later, on October 5, 2021, we made the decision to take her off life support.

Alyssa lived with severe FSHD, but she was never defined by it. She was intelligent, curious, and deeply driven. At the time of her passing, she had written over 165 pages of a book and was reading the DSM-5 out of her own curiosity. She had plans for her life and so much she still wanted to do.

As a caregiver, the hardest part of this disease is the uncertainty about the future. After losing one daughter, there is a constant awareness of how unpredictable this condition can be and how much time we may or may not have. At the same time, I try to give Nessa hope that treatment options will become available and that her future is not already defined by this disease.

What I would most like to see change is access to effective treatment. For our family, this is not theoretical. It is life or death. This disease directly impacts independence, quality of life, and, in our case, survival.

I hold onto hope because I know there are clinical trials and promising research underway, but time matters. For families like ours, this isn't just science moving forward - it's the possibility that my daughter will have the chance to go to college, build a career, live independently, and have a future every parent wants for their child.

What I hope people understand most is that early-onset FSHD is not just a medical condition. It reshapes childhood, adolescence, independence, and family life in ways that are hard to fully describe unless you live it. It affects every part of daily life and every decision about the future.

I already lost one daughter to FSHD. Every day, I wake up determined to make sure I don't lose another child to a disease that science has the potential to change. I can't change what happened to Alyssa, but I hope that the work being done today will give Nessa, and every child like her, the chance to live the life they deserve.

Despite everything, I still hold onto hope for my daughter and for other families like ours. I hope that meaningful treatments arrive in time to change the course of this disease.

IX. Disclaimer

Discussions in Patient Listening Sessions are informal. All opinions, recommendations, and proposals are unofficial and nonbinding on the FDA and all other participants. This report reflects the FSHD Cure Initiative's account of the perspectives of patients and caregivers who participated in the Patient Listening Session with the FDA. To the extent possible, the terms used in this summary to describe specific manifestations of FSHD, health effects and impacts, and treatment experiences reflect those of the participants. This report is not meant to be representative of the views and experiences of the entire early-onset FSHD patient population or any specific group of individuals or entities. There may be experiences that are not mentioned in this report.

X. Contact

Ally Roets

Vice President, FSHD Cure Initiative

ally@fshdcure.org or

info@fshdcure.org

<https://www.curefshdforall.com>

520.507.1209

For more information about the FSHD Cure Initiative and its mission to accelerate access to effective treatments for all individuals with FSHD, please contact the address above.