FSHD Advocate

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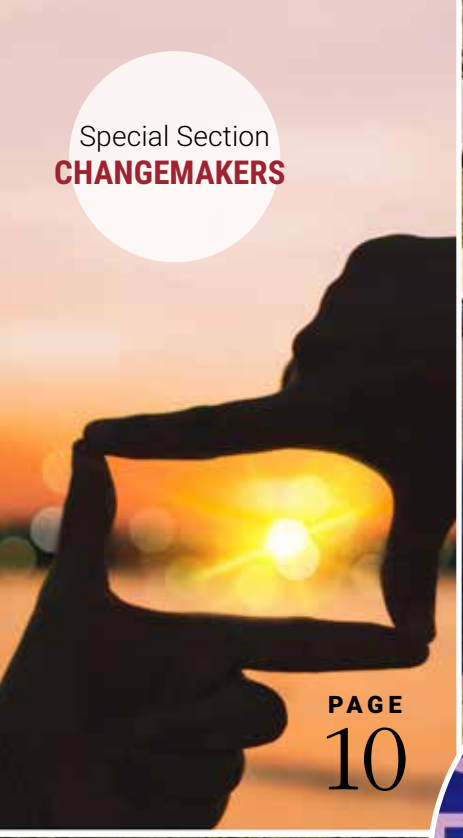
FSHD SOCIETY

Building a better world

IT WILL TAKE ALL OF US



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FSHD Advocate

The FSHD Society is the world's largest research-centered patient advocacy organization focused on facioscapulohumeral muscular dystrophy (FSHD). This genetic condition affects one million people worldwide and causes lifelong, progressive muscle weakness that can result in significant pain and disability. We are accelerating the development of treatments, empowering and activating the FSHD community, and making sure no one has to face this disease alone. The Society does not endorse any of the drugs, procedures, treatments, or products discussed in its reporting. We urge you to consult your physician about any medical interventions.

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June Kinoshita, Editor
june.kinoshita@fshdsociety.org

Look for us on the Internet at *fshdsociety.org*.
We thank the FSHD Society staff for their editorial assistance.

You matter

The profound impact of one person


Mark A. Stone

Our families' stories of life – the simple joys experienced, and, at times, the heartache suffered – have had a profound effect on me. Recently, a recurring theme has seeped into my every conversation and meeting – a story about how one person's encouragement, guidance, or involvement has changed an outlook, attitude, or life for the better.

For instance, Mike told a story of when he was first diagnosed. Uncertain of what the future would hold, he was put in contact with Ashley, who in a matter-of-fact way provided practical, sound advice on travel tips. Years later, Mike is still expressing gratitude for Ashley and paying it forward to have an outsized influence on others.

Or consider Joanne, who was diagnosed earlier this year after a decade-long diagnostic journey. She attended our biennial Connect Conference, learned of a Walk & Roll event in her area, and began reaching out to her friends and neighbors. And did they ever respond! She had more than 100 people on her team and raised more than $100,000. Joanne's astonishing achievement was possible because our volunteer chapter directors provided the platform for her to engage and inspire.

Scores of interactions like this take place every month. Ordinary individuals, without knowing it, make an extraordinary impact, and that impact reverberates to others. Every action, no matter how small, creates ripples. Its effects can spread far beyond the initial interaction – producing life-altering and world-changing results.

While it's true that collective action by an engaged community is often needed for large-scale outcomes, never underestimate the power of a single person to make a profound change on those around you. As Dr. Seuss wrote, "To the world, you may be one person – but to one person, you may be the world."

Every action, no matter how small, creates ripples. Its effects can spread far beyond the initial interaction – producing life-altering and world-changing results.

Mark

Mark Stone
President and CEO
FSHD Society

Editor
June Kinoshita

Copy Editor
Elaine Alibrandi

Graphic Design
Leslie Anne Feagley

FSHD Society
(781) 301-6060
info@fshdsociety.org
fshdsociety.org

To send general mail:
75 North Main Street, Suite 1073
Randolph, MA 02368, USA

To mail donations:
PO Box 411617
Boston, MA 02241-1617, USA

The Global FSHD Innovation Hub

Accelerating clinical trials and access to treatments

BY LAWRENCE KORNGUT, MD, AND BLAINE PENNY, CALGARY, CANADA

This is a very exciting time for the FSHD community, as new therapeutics are advancing through clinical development and the global patient community is coming together to collaborate as part of new initiatives.

This began with the FSHD Society and FSHD Canada Foundation creating Project Mercury two years ago to develop a global framework and toolkit to support the acceleration of clinical trials and ensure patients could get access to new therapies when they are approved.

Blaine Penny and Lawrence Korngut, MD

Over the past year, Lumiio has been working closely with the FSHD Society and FSHD Canada Foundation on the next steps to operationalize this important work. The result is the creation of the Global FSHD Innovation Hub (the Hub) just a few months ago.

The Hub is owned by the FSHD Society, and its mission is to provide a singular global end-to-end offering for biopharma companies to accelerate their products for FSHD and get these potential new treatments to people faster.

The Hub evolved through the global vision and leadership of the FSHD Society, and is a formal partnership among the FSHD Society, TRiNDS, and Lumiio.

Here is what they bring to the table:

- The FSHD Society is focused on patient engagement, education, support and healthcare navigation, and access to clinical trials.
- TRiNDS is a neuromuscular disease (NMD)-specific contract research organization with a successful track record of 30+ projects and a global network of 200+ NMD-experienced sites. TRiNDS will play a key role in site selection and executing the trial operations to improve overall trial capacity and success.
- Lumiio specializes in patient registries, real-world data (RWD) platform technology, site optimization, and the development and validation of digital healthcare tools. (The FSHD Society's BetterLife project was built on the Lumiio platform.)

Why is the Hub needed?

More than 80% of clinical trials experience significant delays due to limitations in sites' capacity for enrolling participants. This becomes a bigger problem as more FSHD trials are launched in the coming years, with increasing demands for qualified sites and recruiting of greater numbers of patients.

As the Hub works toward supporting the readiness of clinical trial sites, it will play a key role in proactively identifying and prequalifying sites that are suitable for FSHD trials and that can recruit patients quickly. Further, the Hub will support these sites to maximize their capacity for enrolling more volunteers into clinical trials.

Site capacity is a global problem, as biopharma companies each need a series of larger clinical trials. In a rare disease like FSHD, this requires trial sites across many countries to develop the evidence needed for regulatory approvals. For example, a Phase 3 trial (the final phase confirmatory trial) could involve more than 15 countries and 50+ trial sites.

Getting a new therapy approved is just the first step of the process for patients to be able to obtain access. Biopharma companies need to go through a rigorous health technology assessment (HTA) to receive reimbursement approval.

Patient data are needed to show that this new therapy provides meaningful value to patients above the

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FDA Patient Listening Session

Upper body mobility: A crucial focus in FSHD treatment

BY ANNA GILMORE, FSHD SOCIETY

On August 23, 2024, the FSHD Society brought a fresh perspective to the Food and Drug Administration's (FDA's) attention during a Patient Listening Session focused on upper body mobility in facioscapulohumeral muscular dystrophy (FSHD). The session aimed to shift the conversation around mobility beyond the ability to walk, and highlight the significant impact of upper body weakness on daily life for FSHD patients.

Most people – even many clinicians – think mobility is only about a person's (in)ability to walk. However, a staggering 96% of people with FSHD experience weakness in the upper arms, shoulders, and trunk. This often-overlooked aspect of the condition profoundly affects a person's ability to do everyday tasks and maintain independence.

The ability to wash your hair, reach to break a fall, or hold an infant is crucial to quality of life. While we would never understate the impact of lower body weakness or loss of ambulation, we felt it was important to shine a light on an aspect of mobility that typically doesn't get as much attention from regulators, payors, or even physicians.

Seven speakers shared their personal experiences by painting a vivid picture of the challenges faced by those living with the condition, and emphasizing the relentless nature of FSHD and the urgent need for treatment options. They gave measured, specific, and quantifiable examples that would resonate with this audience, and answered questions with insight and grace.

The session drew significant attention from the FDA, with more than 40 representatives from 17 offices across three different centers (Drugs, Biologics, and Devices) in attendance. This broad interest underscores the growing recognition of FSHD's impact and the need for effective treatments.

"Having such a large and diverse audience for this meeting was telling," said Mark Stone, FSHD Society CEO. "We know regulators at the FDA – and around the world – are paying attention to what the FSHD community has to say. This conversation will help set the standard for better treatments down the road."

We are grateful to all who participated. Community panelists: Amy Bekier, Maggie Eggleston, Heloise Hoffmann, Evan VanDyke, Jack Gerblick, Debbie Eggleston, and Archer Sverdrup; clinical expert Nicholas Johnson, MD; and advisors and supporters James Valentine, Larry Bauer, and Paul Melmeyer.

We are producing a summary report of the meeting, which will be available publicly on the FSHD Society's website at *fshdsociety.org/our-impact/advocacy/*.

Getting treatments to young patients

MOVE Peds is essential to pave the way

BY NATALIE KATZ, MD, AND KATY DE VALLE, RESEARCH PHYSICAL THERAPIST

More than 50% of people with FSHD develop symptoms as children or teens. When treatments to slow or stop FSHD are approved, common sense suggests that it will be best to treat patients as early as possible to save healthy muscles.

Katy De Valle

Natalie Katz, MD

Yet obstacles stand in the way of reaching this common-sense goal. Most importantly, data are scarce on how symptoms evolve over time in pediatric populations. There have been few studies evaluating the sensitivity of "clinical trial outcome measures," which are methods to measure whether children on treatment are benefiting compared to those who are untreated.

To overcome this challenge, Jeffrey Statland, MD, of the University of Kansas (KUMC), and Ian Woodcock, MD, of Murdoch Children's Research Institute, are co-leading an international effort to better understand disease progression in childhood-onset FSHD.

This prospective natural history study, MOVE Peds, will be similar to the ongoing MOVE (Motor Outcomes to Validate Evaluations) FSHD study, but specifically designed for children.

Initial evaluations will be conducted at seven centers across the US and one in Australia. Additional sites may be added in the future.

What to expect if your child participates

Participants in the study will be seen at six-month intervals for a total of five visits over a 24-month period.

At each visit, participants will complete a variety of assessments including: Strength testing, functional measures (mainly consisting of how quickly they are able to complete certain tasks), breathing tests, collection of biological samples for biomarker development, and surveys to capture different aspects of how FSHD affects their lives.

One of the most exciting things about this study is that children will also be evaluated using Reachable Workspace and whole-body magnetic resonance imaging – the first time these assessments will be done specifically in children with FSHD.

We anticipate the study will begin enrolling toward the end of 2024. Our goal is to recruit at least 80 children with FSHD who are 5 to 18 years old at the time they enroll.

Participants must have genetic confirmation of FSHD as well as clinical symptoms consistent with FSHD to enroll.

Many companies are actively developing treatments for FSHD, and several are interested in making these available to children. We hope MOVE Peds will help make this vision a reality.

Where will the study be done?

IN THE US

- University of Kansas, Kansas City, Kansas
- University of Rochester, Rochester, New York
- Duke University, Raleigh-Durham, North Carolina
- Nationwide Children's Hospital, Columbus, Ohio
- Stanford University, Palo Alto, California
- University of Utah, Salt Lake City, Utah
- University of Iowa, Iowa City, Iowa

IN AUSTRALIA

- Murdoch Children's Research Institute, Melbourne

To enroll your child in MOVE Peds, contact:

IN THE US

- Michaela Walker *(mwalker20@kumc.edu)*
- Leann Lewis *(Leann_Lewis@URMC.Rocester.edu)*
- Rebecca Clay *(rclay@kumc.edu)*

IN AUSTRALIA

- Katy de Valle *(katy.devalle@rch.org.au)*

Beyond REACH

With development of losmapimod halted, what's next?

BY JUNE KINOSHITA, FSHD SOCIETY

In September, we received news that Fulcrum has decided to halt its development of losmapimod. Their decision was based on their analysis of data from the Phase 3 REACH trial.

The results summarized by Fulcrum in its press release contradict findings from multiple previous studies. The REACH data reported small improvements in Reachable Workspace (RWS) not only in people taking the drug but in those taking placebo.

This contradicts data from more than five years of natural history studies of FSHD, which have never shown an improvement in RWS in untreated individuals. Likewise, the Phase 2 ReDUX4 trial of losmapimod showed a decline in the placebo group versus improvement in the treated group. In addition, the Open-Label Extension (OLE) study showed sustained benefits of losmapimod up to 96 weeks in those who were on the drug in ReDUX4, and stable symptoms or improvements for those from the placebo group who began taking losmapimod after the ReDUX4 trial ended.

Over the coming months it will be important to dive deeper into REACH data before broader conclusions can be made. Was the REACH cohort different at baseline (the start of the trial) than the ReDUX4 cohort? In ReDUX4, scores from each participant's dominant arm and non-dominant arm were analyzed separately and showed a larger effect in the non-dominant arm. In contrast, the REACH analysis combined the scores from both arms. What impact did this have on the outcome?

The FSHD Society plans to analyze the data further with Fulcrum and other researchers.

What does this mean for our community?

For patients, it's important to remember that there are many promising therapies in development. Losmapimod reduced *DUX4* through an indirect pathway. Other companies, such as Avidity Biosciences, Dyne Therapeutics, Arrowhead Pharmaceuticals, and miRecule, are pursuing approaches that knock down *DUX4* directly.

Avidity's interim data are highly encouraging, and the next phase of its clinical trials will begin enrolling participants by the end of 2024. A second study is planned for early in 2025.

Other companies with FSHD programs may be concerned about using RWS as a clinical trial endpoint. Studies in FSHD and stroke indicate that it is a valid tool, but the criteria for selecting your test population and how you analyze the data are likely to be critically important. We need further insight on this question.

Fulcrum's decision is a setback, but not a failure. There will be important lessons to learn, and we are closer to treatment because of Fulcrum's work.

Clinical trial snapshot

To learn about clinical trial phases, visit *fshdsociety.org/clinical-trials/*.

For easy-to-understand details about each trial, visit *fshdsociety.org/for-patients-families/clinical-trials/*.

The information shared here is accurate as of October 2024. For status updates on specific locations, search on trial keywords at *clinicaltrials.gov*.

An open-label extension (OLE) is a study in which participants of a placebo-controlled trial are offered the option to continue to be studied after the trial has been completed. All participants in the OLE receive the active drug, so the study is no longer blinded or placebo controlled.

REINFORCE by Centre Hospitalier Universitaire de Nice, principal investigator Sabrina Sacconi, funded by Hoffmann-La Roche

QUICK FACTS	
Drug	Satralizumab
How Is It Given?	Injection under skin
Phase	2
Participants	40
Placebo	Yes
Genetic Testing	Required
Rx Duration	Double-blind phase, at weeks 0, 2, 4, and every 4 weeks thereafter for 48 weeks; open-label phase, same dosing for 48 weeks + follow-ups; total 116 weeks
Study Visits	~16
Notable Activities	MRI
Open-Label Extension	Yes

STATUS	
Enrollment	Began January 2024
Data Expected	Mid-2026
Locations	Ottawa, Canada; Nice, France
Learn More	*clinicaltrials.gov/study/NCT06222827*

WHO CAN PARTICIPATE?

- Age 18-65
- FSHD1
- Ricci score 2-4, able to walk without support
- Must be able to do MRI

FORTITUDE sponsored by Avidity Biosciences

QUICK FACTS	
Drug	Del-brax (AOC 1020)
How Is It Given?	Intravenous infusion
Phase	1/2
Participants	72
Placebo	Yes, 2:1
Genetic Testing	Required, provided by study
Rx Duration	8 doses
Study Visits	~20, some may be virtual
Notable Activities	MRI, leg muscle biopsy
Open-Label Extension	Yes

STATUS	
Enrollment	Currently enrolling
Data Expected	Second half of 2026
Locations	US, Canada, UK
Learn More	*fortitude-study.com* *clinicaltrials.gov/study/NCT05747924* *fshdsociety.org/avidity-fortitude-trial/*

WHO CAN PARTICIPATE?

- Age 16-70
- FSHD1 or FSHD2
- FSHD clinical score of 2-14
- Able to walk 10 meters without assistance
- Reachable Workspace score
- Must have leg muscle suitable for biopsy and be able to do MRI

ARO-DUX4 trial sponsored by Arrowhead Pharmaceuticals

QUICK FACTS	
Drug	ARO-DUX4
How Is It Given?	Intravenous infusion
Phase	1/2a
Participants	60
Placebo	Yes, 3:1
Genetic Testing	Required, provided if needed
Rx Duration	Part 1: duration 3 months Part 2: 4 doses over 1 year
Study Visits	~20
Notable Activities	MRI, leg muscle biopsy
Open-Label Extension	Yes, if warranted by study results

WHO CAN PARTICIPATE?

- Age 18-70
- FSHD1
- Clinical Severity Scale 3-8
- Must have leg muscle suitable for biopsy and be able to do MRI

STATUS	
Enrollment	Currently enrolling
Data Expected	TBD
Locations	New Zealand, Australia, Canada, Europe
Learn More	*fshdsociety.org/arrowhead-trial* *clinicaltrials.gov/study/NCT06131983*

MANOEUVRE sponsored by Hoffmann-La Roche

QUICK FACTS	
Drug	GYM329 (aka RO7204239)
How Is It Given?	Injection under skin
Phase	2
Participants	48
Placebo	Yes, 1:1
Genetic Testing	Required, talk to your local site
Rx Duration	Every 4 weeks for 52 weeks
Study Visits	At least every 4 weeks
Notable Activities	Wearable device, MRI
Open-Label Extension	Yes, for 52 weeks

WHO CAN PARTICIPATE?

- Age 18-65
- FSHD1 or FSHD2
- Ricci score: ≥ 2.5 and ≤ 4 (must be able to walk unassisted)
- Must be able to do MRI

STATUS	
Enrollment	Complete. Study is active.
Data Expected	Q3 2026
Locations	US, Denmark, Italy, UK
Learn More	*forpatients.roche.com* *clinicaltrials.gov/study/NCT05548556fshdsociety.org/roche-manoeuvre-trial/*

What you can do to be trial-ready

These steps will reduce delays and improve your chances of enrolling in a trial. You will also need to meet the eligibility criteria for the specific trial. There's no guarantee, as many factors must be considered in selecting people for a trial, but these measures can help.

- Make sure you have been diagnosed by a doctor.
- Get a genetic test confirming FSHD.
- Become a patient at an FSHD Clinical Trial Research Network location.
- Enroll in the MOVE or MOVE+ study.
- See your neurologist every year.
- Follow exercise recommendations and be as healthy as possible.
- Sign up to get FSHD Society email alerts.

Learn more:

Building a better world

It will take all of us

JUNE KINOSHITA AND ERIN SAXON, FSHD SOCIETY

When the Society was formed in 1991, no one knew the cause of the condition. People with FSHD had no organization to mobilize them to action.

This did not stop Daniel Perez and Steve Jacobsen from trying. They focused on these goals: Find the gene for FSHD. Recruit a scientist, then another, to help. Find doctors who see FSHD patients. Ask patients to get involved.

The Society built itself up one person at a time, laying one brick after another. We built a path that grew into a lane, then a highway, and now a beltway that encircles the globe. This infrastructure is speeding up drug development and empowering patients everywhere.

A new generation is taking up the work. Here are the stories of some of these changemakers. They give us hope and should inspire each of us to join them in building a better future.

ARCHER SVERDRUP

Building an early-onset community

Meet Archer Sverdrup, a St. Louis native with a love for power soccer, arts and crafts, and hanging out with their loyal German Shepherd, Blue.

Archer recently joined a power chair soccer team, where they enjoy the thrill of the game and the camaraderie it brings.

Living with symptoms of FSHD since birth, Archer has navigated a long road to diagnosis and self-acceptance. Now, they're channeling their experiences into creating meaningful connections for others.

Archer Sverdrup and their dog, Blue

Recently, Archer co-founded Everything Early Onset, a support group specifically for people who are living with early-onset FSHD.

"There's a very particular grief that comes with losing things you never had," Archer shares. Their group has quickly become a safe space for those with similar experiences, where members can exchange insights and feel seen.

Looking to the future, Archer plans to pursue a doctorate in occupational therapy, aiming to help others with disabilities in impactful ways. Through their community work and advocacy, they're already creating change in the FSHD community.

MARISA SPAIN
Advocacy with humor and hacks

Marisa Spain has a knack for making everyday advocacy both fun and impactful. Based in southeastern Michigan, Marisa brings fresh energy to accessibility awareness with her unique YouTube channel, Disabled in Nature, where she reviews public restrooms for accessibility in a series called "When Nature Calls." Her witty take on something as essential as restroom accessibility adds humor to the serious gaps she's out to close.

"You don't know how important restroom access is until it's denied to you," Marisa says, explaining her mission to help both business owners and viewers understand what true accessibility looks like.

In addition to her work online, Marisa also co-leads the Everything Early Onset Zoom group alongside Archer Sverdrup, creating a safe, open space for people of all ages who were diagnosed with FSHD early in life.

The group meets monthly, focusing on shared experiences and life hacks to navigate FSHD. "I hope our group provides a sense of community – something I didn't have much of when I was younger," Marisa shares. For her, this group is about more than support; it's a way to let others know they aren't alone.

Marisa's passion for advocacy and community building is part of who she is. "The best thing we can do for each other right now is to build a strong community," she reflects, acknowledging how the group meets both practical needs and the emotional support crucial for living with FSHD.

When she's not working on her YouTube channel or helping to lead the early-onset group, Marisa enjoys life in Michigan with her husband and their dog, Harley. Through her work and her advocacy, Marisa shows how powerful a positive, practical approach to challenges can be.

Marisa Spain

Justin and Amanda Hill

AMANDA HILL
A personal mission to advance research

Amanda Hill, a Colorado native, lives with her husband, Justin, and their two dogs in the Denver area. Together, they share a love for travel, great food, and discovering new spots in their local dining scene.

Professionally, Amanda has built a career in biomedical research, where her knack for project management led her to roles overseeing clinical studies and supporting collaborative research efforts.

After Justin's FSHD diagnosis, Amanda sought out the FSHD Society and soon became a chapter leader for Colorado, where she helped connect local families and build a supportive community. "Being a chapter leader gave me the chance to connect with people in the same boat," Amanda shares.

Now, as the Society's senior director of research and care, Amanda plays an integral role in projects like the BetterLife FSHD platform. "We're still in the early days, learning the system and building out its potential, but the response has been incredible," she says, excited about the impact of BetterLife in connecting patient data to research.

With her mix of research expertise and personal commitment, Amanda is dedicated to driving meaningful progress for the FSHD community.

MICHELLE MELLION, MD
Finding a way around all obstacles

One lesson Michelle Mellion, MD, absorbed from seeing patients at a Muscular Dystrophy Association (MDA) neuromuscular clinic is that people with FSHD "are great adapters, using what they can and taking what they have learned about themselves and FSHD to overcome any challenges to accomplish their goals."

A neurologist and faculty member at Brown University's Warren Alpert Medical School at the time, Mellion says the patients have inspired her to this day as she navigates through multiple roles in academia and biopharma companies.

After earning her undergraduate degree in molecular biology at Colgate University, Mellion attended Wake Forest University Medical School for her MD and completed her internship in medicine, residency in neurology, and fellowship in clinical neurophysiology at Brown University. At Brown, she spent more than a decade teaching, conducting research, and seeing patients.

After transitioning to industry, with roles at Biogen and Vertex, Mellion led development of the FSHD program at Fulcrum Therapeutics through Phase 2 clinical trials. Currently, as chief medical officer at PepGen, Mellion leads the development of oligonucleotide therapeutics for rare neuromuscular diseases.

Even while working in industry, Mellion continued to serve the neuromuscular community as an attending physician in the MDA clinic at Brown and then at the pediatric neurology clinic affiliated with Tufts University. She continues to make significant contributions to FSHD through her role as medical advisor to the FSHD Society.

Michelle Mellion, MD

The path to treatments has not been smooth. "We may hit a couple of bumps along the way," she notes, "but just as people living with FSHD do, we researchers will also have to take the time to examine what we have learned.

"People living with FSHD do not give up. If there is not a straight path forward, they will find a way around," she says. "This community inspires me to do my best every day."

Chelsea Moeller with her daughter, Charlie, and husband, Travis.

CHELSEA MOELLER
Speaking honestly

Originally from the Midwest, Chelsea Moeller now calls Maine home, where she lives with her husband, daughter, and their 125-pound Newfoundland, Del. The close-knit community of her town makes it an ideal place for her family.

Chelsea also shares a deeply personal connection to FSHD, navigating life with the condition herself. Her journey inspired her to create HonestlyFSHD, a blog where she reflects on her experience with openness and authenticity. "I needed a place where I could be honest about my reality and educate the people around me who could see the story unfolding," she reflects, creating a space for others on a similar path to find connection and understanding.

Professionally, Chelsea has built a robust career in nonprofit development, holding roles focused on creating impact for nearly two decades. Most recently, she served as the vice president of advancement for a private college in Maine, following her role as director of donor engagement and capital projects at the Children's Center in Augusta.

Now, as director of development at the FSHD Society, Chelsea sees her work as more than fundraising. "I don't think of it as asking for money," she explains. "I see it as connecting people who genuinely want to make a difference with the lives they can change."

BILL SARRAILLE

Putting a lifetime of lawyering to work for our community

"I couldn't do a chin-up to save my life," Bill Sarraille recalls of his third-grade self. This did not deter him from sports, but he was plagued by mysterious pains in his back, hip, and knees.

Then, when he was in law school, he was unable to pivot in the middle of a football game. "The next morning, I got out of bed and looked at my right lower leg, and half of it was gone." The muscle had deteriorated, almost overnight.

After visits to many doctors, he finally met one who said he had FSHD or a similar muscular dystrophy. "Nothing can be done," the doctor told him, "but try to live a stress-free life."

That wasn't going to happen in Sarraille's chosen career in law. "So I didn't do anything related to my FSHD for years and years. I just put my head down and worked."

After his diagnosis, his mother and two sisters were also diagnosed with FSHD. His sisters' disease progressed faster than his. His eldest sister was bedridden for 10 years before she died, and his other sister is now bedridden as well.

"It's very sad to see other family members affected even more severely than I was," he says. "In my career as a healthcare lawyer, that family perspective loomed over what I did."

Recently retired, Sarraille had specialized in healthcare regulatory and compliance matters for more than 30 years. His expertise in launching rare disease treatments and navigating coverage and reimbursement hurdles is invaluable now in his role as a board member and advisor to the FSHD Society.

"Regulators have to condition themselves to separate emotionally from what is very real to us," he says.

Bill Sarraille with his sister, Mary Jean, at his graduation from Harvard Law School.

"It's an old plaintiff lawyer's axiom that the job of a plaintiff in a trial is to show their wounds to the jury.

"It's going to be up to us to show our wounds. To show that our suffering, our families' suffering, deserves attention, deserves treatment, and that we aren't going to settle for an answer that doesn't provide meaningful access to therapy."

CAROLYN LLOYD

Connecting generosity with purpose

Carolyn Lloyd's journey as chief development officer of the FSHD Society reflects her lifelong commitment to service and empathy for those affected by chronic illness. Her role as a caregiver for her mother, who had multiple sclerosis, taught her firsthand about the complexities of chronic conditions.

"I understand the challenges not just for the individual, but for the whole family," she says, a perspective that grounds her work.

Her career in development has spanned healthcare, higher education, and rare disease, giving her a rich foundation of experience.

"I've always been intentional about learning every aspect of development to understand how to make the biggest impact," Carolyn reflects.

Carolyn Lloyd

Each step along her career path – from events and annual funding to corporate relations – has deepened her skill set and passion for supporting families.

For Carolyn, development is about connecting people's generosity with opportunities to create change. "It's about fulfilling people's desire to help," she says.

Inspired by the FSHD community's strength and support for one another, Carolyn strives to make sure no one feels alone on their journey. "Being around these families is incredibly moving," she says. "I want everyone who supports this cause to know they're part of that."

Now based in Kansas City with her husband, Carolyn enjoys live music, especially jazz, along with hiking, playing the piano, and creating art.

LUCIENNE RONCO, PHD
Science with a heart

Lucienne Ronco's journey is a testament to the power of curiosity and compassion. With roots in the Pocono Mountains of Pennsylvania, she embarked on a path that would lead her to the forefront of rare disease research and drug discovery. Her academic quest took her from Tufts University to the University of California, Los Angeles, where she earned her PhD in biological chemistry before returning to the East Coast for postdoctoral training at Harvard Medical School.

Lucienne Ronco, PhD

Throughout her career, Ronco has navigated the complex landscapes of both pharmaceutical giants and biotech start-ups, rising to leadership positions that have shaped the direction of drug development. At AstraZeneca she ascended to the role of global director of translational sciences, where she honed her skills in bridging the gap between laboratory discoveries and clinical applications.

It was during her tenure at the Massachusetts Institute of Technology-Harvard Broad Institute that Ronco's passion for rare diseases truly took hold. Here, she witnessed firsthand the profound impact that identifying genetic mutations could have on therapeutic success.

This experience would prove pivotal when the opportunity arose to join Fulcrum Therapeutics, a decision that would immerse her deeply in the world of FSHD research. At Fulcrum, Ronco found herself surrounded by some of the brightest minds in science, united in their quest to unravel the mysteries of facioscapulohumeral muscular dystrophy.

Despite the termination of Fulcrum's FSHD program, Ronco remains optimistic. "The therapeutic community has generated so many ways to knock down a protein, so we have many possible approaches to try," she says.

Now, as chief science officer of the FSHD Society, Ronco brings a wealth of experience and a burning desire to give back to the community. She approaches her work with the same empathy and gratitude that she developed during her years at Fulcrum, when she was moved by the generosity and courage of FSHD patients. "They gave us biopsies. They gave us MRIs. All this while overcoming mobility challenges just to get to the study sites," she recalls.

Ronco's commitment to community plays out in other ways. She serves in her local government in Wellesley, Massachusetts, and has volunteered to teach STEM to middle and high school students in Boston. When not advancing the frontiers of medical science, she enjoys hikes in the New England wilderness and travel to distant lands.

Lucienne Ronco's quest for scientific knowledge, tempered by deep empathy for those affected by rare diseases, has brought her to the FSHD Society. Here, she bridges the esoteric world of molecular biology and the human quest for hope and healing.

DON BURKE
A passion for adventure and advocacy

Don Burke recently completed a remarkable 93-day road trip, covering 28,000 miles across 36 states and four Canadian provinces in his electric vehicle. This journey is part of why he decided to retire early. "This trip was a bucket list adventure," he said, "and it reminded me just how much is still out there to see."

Don's connection to FSHD runs deep. His family is affected and has known about the disease since the 1970s. "I've always leaned into it, never denied or ignored it," he said. He contributed to foundational efforts for the FSHD Society, attended the first support group meetings, participated in clinical trials, and even helped build the Wikipedia page on FSHD.

Don continues to adapt and push boundaries. "The sense of adventure never leaves," he said. Through his work with the FSHD Society's Patient Engagement Committee and his commitment to renewable energy, Don's passion for advocacy shines in every aspect of his life.

Don Burke and his electric vehicle atop Mt. Blue Sky in Colorado.

Romantic comedy puts FSHD in the spotlight

Showing what "Hollywood can do when they get behind us"

BY RICH HOLMES, SANDWICH, MASSACHUSETTS

When was the last time you went to the movies and saw a film starring an actor who has muscular dystrophy?

Never, right?

That changed on August 15 when the independent film *Good Bad Things* opened in AMC theaters across the US. The MDA (Muscular Dystrophy Association) took the unusual step of promoting the opening, and sold tickets to see the movie on the streaming platform VEEPS in late September.

"I got very emotional. It was the first time I saw a film on the big screen starring someone who resembles me," said Mindy Henderson, MDA vice president of disability outreach and empowerment, and editor-in-chief of the MDA's Quest Media.

The movie could be described as a romcom with depth. It stars Danny Kurtzman as Danny, a 30-something guy who, with his best friend, housemate, and business partner, Jason, runs a struggling web design and marketing outfit.

Danny hopes to land a big account to put his business back in the black, and shoots for a contract to rebrand an established dating app. With the encouragement of Jason, he ends up meeting someone on the app. Henderson praised the role of Danny for going beyond stereotypes of disabled people.

"He's also an entrepreneur, he's a friend, he's a boyfriend," she said.

Kurtzman has FSHD (facioscapulohumeral muscular dystrophy), one of the three most common forms of muscular dystrophy. He's severely affected, unable to walk, and relies upon a scooter to get around. The camera doesn't shy away from showing his thin and bony body, but welcomes us to embrace it, especially in a scene with Danny's romantic interest, Madi (Jessica Parker Kennedy), a photographer.

"The photo shoot – she captured Danny in so many beautiful ways," Henderson said.

The love story includes the deep friendship Danny has with Jason, which draws upon the real-life friendship of Kurtzman and Shane D. Stanger, the film's co-writer and director.

"The on-screen chemistry … with Brett (Dier, who plays Jason)" was filmed "while my best friend was behind the camera," Kurtzman said.

The film is Stanger's first feature-length film and Kurtzman's first acting role. Kurtzman contributed to writing the film. The film mines much of Kurtzman's life, adding to its authenticity; he's an entrepreneur with experience in retail and fashion.

Henderson said other groups have gradually won representation in film and entertainment media. Now it's disabled people's turn.

"Disability is kind of the last man standing," she said.

The movie's message is that love is worth the risk of failure, and reluctance to try is itself a sort of disability, and makes the film relatable to the general public.

"I think we all get in our own way," Kurtzman said.

It's been two years since shooting began, and the film has won awards at several festivals. Now, the MDA and the group behind the movie hope to sell Hollywood on the viability of this sort of film.

"There is an appetite and demand for this kind of film," Henderson said, adding that she would like to see "more and more films made by and starring people with disabilities."

Good Bad Things will "show what Hollywood can really do when they get behind us," Kurtzman said.

Editor's note: Article reprinted (with editing) courtesy of *Cape Cod Health News*.

BetterLife FSHD

An innovative platform designed to give you tools to manage your health while contributing to FSHD research

Take control of *your* FSHD story.

PRIVACY.FLOWCODE.COM

www.BetterLifeFSHD.Org

BetterLife FSHD

Customized Health Insights
Secure Data Contribution
Clinical Trial Matching

Our mission is to provide you with the tools and resources to better understand and manage your FSHD, all while contributing to groundbreaking research.

Participation in BetterLife FSHD includes:

Symptom Tracking: Monitor your health and track symptoms over time, gaining insights into your personal health journey.

Customized Resources: Access a wealth of information and support tailored specifically to your needs and symptoms.

Clinical Trial Matching: Discover research studies and clinical trials for which you may be eligible, opening doors to new treatment opportunities.

Secure Data Contribution: Safely share your health data with approved researchers, knowing your privacy is protected.

By completing participant surveys, your voice contributes to a larger understanding of FSHD, helping researchers and healthcare professionals develop more effective treatments.

You're not just managing your FSHD:
you're paving the way for a better future for everyone affected.

FSHD SOCIETY

Fundraise YOUR way: Do-It-Yourself (DIY) Fundraising

At the FSHD Society, we know that every advance is made possible by the incredible support of people like you. One way you can join the fight is through our DIY fundraising program, which allows you to create events that are as unique and creative as you are. Whether it's a golf tournament, a bake sale, or a personal challenge, your event can make a huge impact.

Our DIY fundraising program is designed to give you the freedom to raise funds in ways that align with your interests, talents, and passions. Whether you are an avid runner, a skilled chef, or simply want to rally your community together, there are countless ways to contribute to the cause. We provide the support and resources you need, while you bring creativity and drive!

The FSHD Society's Anna Gilmore and Kim Shanley.

Fundraise Your Way!

DIY events from our community

Here are just a few examples of DIY events that have already made a difference.

- **Pampered Chef Event.** Sandra in Alabama turned her love of cooking into a fundraising powerhouse. By donating a portion of sales from her event, this creative fundraiser made a helpful contribution to the FSHD Society.
- **Golf Tournament.** Steve, a golf enthusiast in Texas, organized his third annual golf tournament to benefit the FSHD Society, bringing together a full day of friendly competition for a good cause.
- **City2Surf Sydney, Australia Run.** Sophie, during her semester abroad, ran the City2Surf race in Sydney, one of the biggest running events in the world, and this year it became a platform to raise awareness and funds for the FSHD Society.
- **Bar Mitzvah Fun Run.** Jonah organized a fun run as part of his Bar Mitzvah project, inviting family, friends, and their community to join in and support the FSHD Society. His event was a heartfelt and creative way to mark this important occasion while contributing to a cause close to his heart.

Start your own DIY fundraiser today

If you've been inspired by these stories, now is the perfect time to start planning your own event. Whether you want to plan something small or big, we are here to help. You can start by visiting our website today.

– Kim Shanley

2024 Walk & Roll to Cure FSHD: Excitement is in the air, and on the trails

More than a baker's dozen of enthusiastic participants joined team Raising the Dough for the New England Walk & Roll on September 22 in Brookline, Massachusetts. Decked out in chef's aprons, "We did not loaf around, but walked, rolled, biked, and rigged around the park," quipped team leader Kathy Senecal. "Walk & Rolls are proof that combining the right blend of ingredients will give rise to success!"

The New England event was one of nearly 30 across the US and Canada this fall. The FSHD Society's flagship grassroots fundraiser hosted 1,900+ participants and 160+ teams toward their goal of raising $1.2 million.

Now in its seventh year, the Walk & Roll has an impact extending far beyond the numbers. It's about community, hope, and empowerment. For people living with FSHD, what can be a lonely journey becomes a mass movement. When that first-ever treatment crosses the finish line, we know that we've played a critical role in making that happen.

– Beth Johnston

Kathy Senecal and her Team "Raising the Dough" at the New England Walk & Roll to Cure FSHD.

WEBINARS

Please register in advance to get the link.

November 21: ReSolve – what we have learned
December 19: Annual drug development update
January 16: Mindfulness for chronic stress
February 20: Anesthesia during surgery
March 20: Physical therapy for overworked muscles

FSHD UNIVERSITY

FSHD SOCIETY RADIO

On the second and fourth Tuesday of every month, listen for a new episode of FSHD Straight Talk with Tim Hollenback. These episodes highlight members of our community who are living rich, full lives and diving into their experiences with FSHD. The podcasts are available to stream on-demand on major streaming platforms.

CHAPTER MEETINGS

Our 30+ chapters are busy planning meetings for this fall, so please visit our Events Calendar for updates. Many meetings are virtual or hybrid. All are welcome to join from anywhere. Please pay attention to the time zones.

THE GATHERING PLACE

All of the Zoom groups listed below meet on a regular monthly schedule. For details about topics and speakers, consult the Events Calendar. To join, register once and you'll receive the link and monthly reminders.

CarePartner Hour
Last Tuesday of every month at 8 p.m. ET | 7 p.m. CT | 6 p.m. MT | 5 p.m. PT

Everything Early Onset
First Friday of every month at 8 p.m. ET | 7 p.m. CT | 6 p.m. MT | 5 p.m. PT

Feeling Fit with FSHD
Second and fourth Thursday of every month (unless a major holiday) at noon ET | 11 a.m. CT | 10 a.m. MT | 9 a.m. PT | 6 p.m. CET| 5 p.m. GMT

Parents' Roundtable
Third Tuesday of every month at 8 p.m. ET | 7 p.m. CT | 6 p.m. MT | 5 p.m. PT

Wellness Hour
Second Monday of every month at 5 p.m. ET | 4 p.m. CT | 3 p.m. MT | 2 p.m. PT

Women on Wellness
First Wednesday of every month at 5 p.m. ET | 4 p.m. CT | 3 p.m. MT | 2 p.m. PT

Young Adults
A Zoom community for empowering young adults impacted by FSHD. Meeting time has changed to the third Monday of every month at 8 p.m. ET | 7 p.m. CT | 6 p.m. MT | 5 p.m. PT

Visit our Events Calendar for updates and to register for events at *fshdsociety.org/fshd-events-calendar.*

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The Global FSHD Innovation Hub

current standard of care to receive reimbursement.

Our goals

One of the primary goals of the Hub is to proactively support existing patient registries and establish new ones such that the data required (e.g., quality of life, activities of daily living, etc.) for an HTA are being generated so patients can access these new therapies as quickly as possible.

Another key function of the Hub is the ability to develop and validate *innovations* that could help diagnosis, mobility, muscle regeneration, finding patient resources, etc., to improve patient outcomes and quality of life.

Some examples of this include use of AI to identify undiagnosed patients through electronic health records, exoskeletons to help with mobility/function, and interactive tools for patients to find relevant helpful information to support their journey and needs (e.g., finding clinicians, clinical trials, equipment, etc.).

Over the past few months, we have engaged in several discussions with biopharma companies, receiving highly positive feedback that affirms our model as an innovative approach for advancing therapies and outcomes in rare diseases.

The authors: Lawrence Korngut is associate professor at the University of Calgary Cumming School of Medicine and leads the University of Calgary Neuromuscular Clinical Trial Program, which has conducted more than 80 clinical trials of new therapies. He is also chief medical officer, and Blaine Penny is director of Innovation and Real Data/Evidence, of the Global FSHD Innovation Hub.

FSHD Advocate

FSHD Society
75 North Main Street
Suite 1073
Randolph, MA 02368

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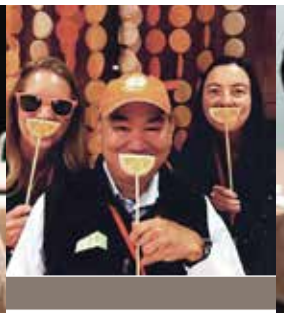


THE FUTURE IS NOW

Together, we passionately strive for a future without the impact of FSHD while ensuring that those affected have the support and resources to live fully today.

We need your continued kindness to press forward. Each donation is more than dollars — it's progress, hope, and confidence in what tomorrow will bring. And there is much to be hopeful for.

Give by December 31, 2024, and double your impact. Generous individuals are matching all donations made by year end. Join us in changing the world for the FSHD community!

FSHD SOCIETY
FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY