# FSHD Advocate

2020 • ISSUE 2

FSHD SOCIETY
LIGHTING THE WAY TO A CURE


# FORWARD TOGETHER

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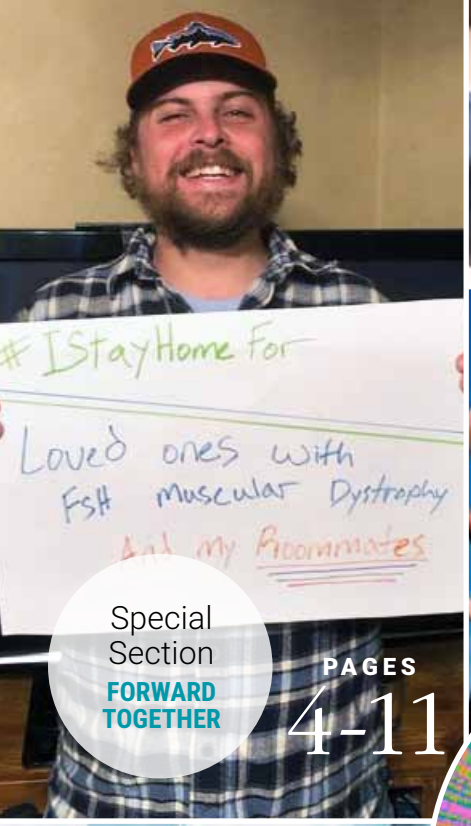


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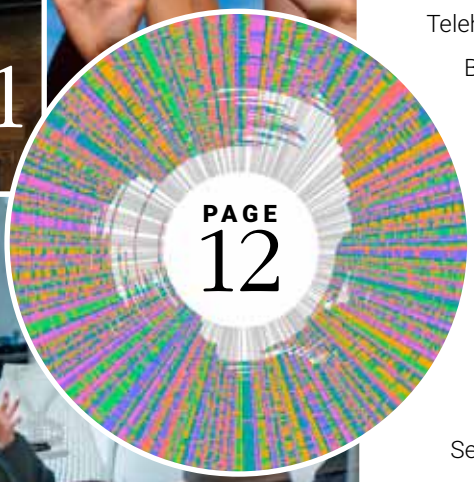

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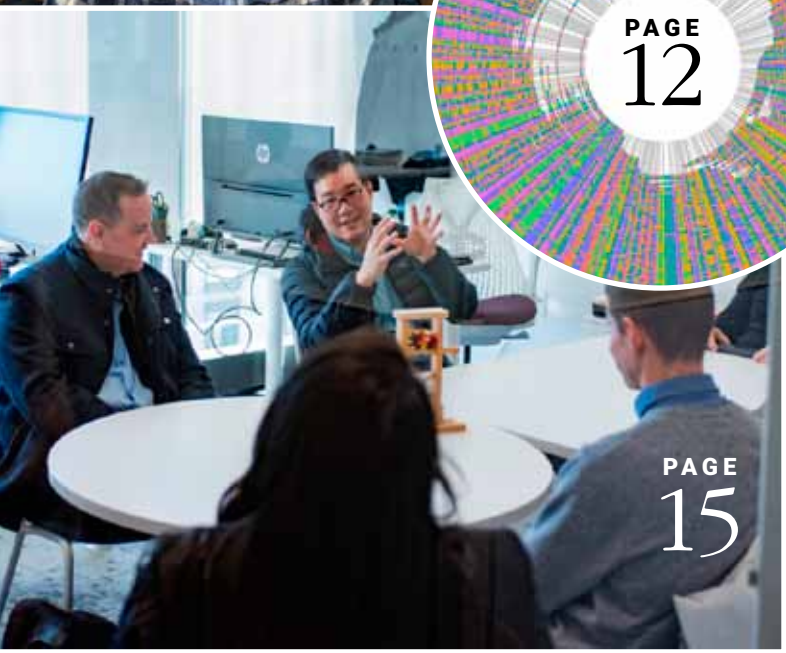

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Our reporting on developments regarding FSHD does not imply that the FSHD Society endorses any of the drugs, procedures, treatments, or products discussed. We urge you to consult your physician about any medical interventions.

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CEO Letter

# Forward Together

*The power of an active community*

While catchphrases have proliferated during the COVID-19 pandemic, they are nonetheless a clarion reminder from public officials, medical professionals, and leaders that we are indeed in this together – and we will get through it together.

We have seen the power of community as residents in cities large and small join together from balconies, fire escapes, and roadsides to honor front-line workers. Expressions of appreciation for delivery personnel and grocery clerks, who have kept our stores stocked and allowed us the privilege of security, have become regular features on news outlets.

The former Secretary of Agriculture Tom Vilsack once said, "People working together in a strong community with a shared goal and a common purpose can make the impossible possible." This aptly describes not only our current situation, but also the power and potential of the FSHD community.

As you read this edition – our first digital-only magazine – you will see that, through innovation, resilience, and commitment to our common goal, the FSHD Society community is more connected and truly stronger together.

Our communal strength is highlighted in Dr. Jeff Statland's article on the ReSolve study – the largest global observational study documenting the effects of FSHD.

**Mark A. Stone**

You will also see it in our signature Walk & Roll to Cure FSHD – now a nationwide, virtual event on September 12 – as our community has expanded with more families joining us in our quest for treatments and a cure for FSHD. And the power and potential of our collective force is certainly evident in the articles about our response to the COVID-19 crisis, and the amazing growth and impact of our national chapter initiative.

The strength of our community lies in the commitment of the individuals within it. We salute those who step forward to become an even bigger part of the solution – people like our volunteer chapter directors and individual fundraising leaders, who reached out into their personal communities to enlarge and empower ours.

We join together from our figurative "balconies, fire escapes, and roadsides" to applaud all of you who work tirelessly and give generously for the day when the impossible becomes possible.

Until then – forward together!

Mark Stone
*President and CEO*
*FSHD Society*



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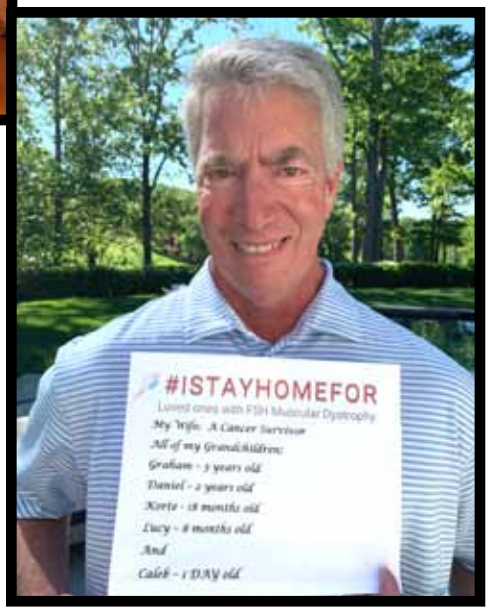
# THE PANDEMIC PIVOT

BY JUNE KINOSHITA, FSHD SOCIETY

When the world shut down in mid-March to stall the spread of COVID-19, the FSHD Society team not only pivoted, but it embraced the challenges posed by the crisis by turning this sow's ear into a silk face mask. As the FSHD Society is a rare-disease organization, many of our stakeholders are accustomed to being socially distanced. The digital highways and byways have been the primary channels where we connect and support one another. Adapting to social distancing through technology was already engrained in the FSHD community.

Our nimble FSHD Society team leaped into action. We developed a rich online program with research updates, mindfulness sessions, chapter meetings, fitness, and more. Many in the community offered help and advice, compiled lists of resources, shared tips and tricks, and reminded us all to attend to our mental well-being. We called these activities "Sequester Camp," injecting a little cheeky fun as our way of defying the darkness of the times and affirming the bonds that make us strong and positive.






The doctors and researchers of the FSHD Clinical Trial Research Network have begun to offer telehealth consultations while working tirelessly to adapt their ongoing projects and even come up with new studies to understand how this pandemic and the public health policies are affecting our community. We worked with the CTRN to develop and distribute a survey on the impact of COVID-19, and hundreds of you responded. Amazing!

Our major conferences this year – the International Research Congress, FSHD Connect, and Voice of the Patient Forum – were forced to go virtual. Even though we were disappointed at our inability to convene in person, we found a silver lining: Anyone with a laptop




or smartphone and decent WiFi could attend. We reached thousands of people with each webinar and radio show. Wonderfully, our meeting sponsors and foundations stuck by us. Financial support from the National Institutes of Health, Bionano, Fulcrum, Association Française contre les Myopathies, Allard, Avidity, Dyne, Genomic Vision, the University of Nevada Reno, Muscular Dystrophy Association, miRecule, PerkinElmer Genomics, Optum Health, and the James Chin Sr. Scholarship Fund made these events possible. We are deeply grateful to them all.

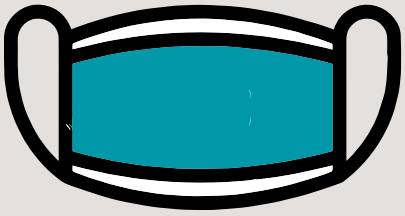
Being forced to think outside the box has pushed us to innovate. We have found new, exciting opportunities that will continue to shape and elevate how the FSHD Society serves the community in years to come. We can't wait to share them with you.

Through all of this, we have deeply appreciated your support and encouragement. Amazingly, in a season when many donor-supported organizations are struggling, our spring campaign raised more than $200,000, a record that testifies to your determination and commitment not to let anything – not FSHD, not COVID-19 – stand in our way. Together, we will prevail.

## COVID-19 advisory

For most individuals with FSHD, the risk of serious illness from COVID-19 infection is the same as for the general population. The CDC and MDA websites provide detailed information about who is most at risk and how to avoid getting infected.

- Older adults (more than 50 years of age), especially those with chronic illnesses such as heart disease, diabetes, compromised immune systems, and lung disease, have the highest risk of serious illness.
- If you have FSHD-associated restrictive lung disease, you should consider yourself at higher risk for serious illness regardless of your age and take extra precautions to avoid becoming infected. This applies to anyone who is using nighttime ventilation or is at or below 50 percent of the predicted value for forced vital capacity (FVC), maximal inspiratory pressure, and peak cough flow.
- If you get sick, inform your care providers that you have restrictive pulmonary disease and should not receive oxygenation without ventilation (e.g., BiPAP or NVS), as this can cause you to retain carbon dioxide and stop breathing.
- Download and carry our Medic Alert Card to warn emergency medical care providers that you have FSHD.



# Adapting nimbly to the new normal

**BY JAMSHID ARJOMAND, PHD, CHIEF SCIENCE OFFICER, FSHD SOCIETY**

"Unprecedented times," "social distancing," "Zoom calls," "new normal…." These phrases have become all too common in our daily, self-sequestered lives and reflect our inherent abilities to adapt to new conditions, no matter how inconvenient or taxing they may be. However, the key drivers at the core of our adaptability are really our skills at prioritization and creativity, combined with a good dose of resilience.

The team at the FSHD Society is no exception and has been adapting quickly to ensure that our research initiatives continue forward. These qualities are also demonstrated by scientific and clinical researchers in the field, who undoubtedly draw their inspiration from the FSHD community at large.

Like most organizations, we ended 2019 by drafting a set of ambitious goals and priorities to advance the FSHD Society's Therapeutic Accelerator program. These plans included a series of workshops, academic and industry meetings, grants, and international conferences peppered throughout the 2020 calendar. Unfortunately, these plans quickly ran aground with COVID-19.

As the pandemic spread across Europe and the US, researchers and clinicians began contacting us about travel restrictions being imposed by their institutions. Faced with the unknown, we initially thought to postpone the upcoming activities, but as the reality of social distancing extended beyond the first month, we went back to the drawing board to reevaluate our plans. One thing we knew from the outset was that stopping would never be an option. With that mindset, we looked to reprioritize and adapt, using new virtual tools as well as input and ideas from our colleagues.

One of our highest-priority meetings this year, the Voice of the Patient Forum (VOPF), was scheduled for April 21 – right in the middle of the shutdown. This was the FSHD community's opportunity to educate the Food & Drug Administration (FDA) on how FSHD is impacting the lives of patients and their loved ones. With FDA staff barred from attending public gatherings in April, we rescheduled for an in-person event on June 29. However, as we remained concerned about public safety, we asked the FDA if they would consider a virtual meeting. To our delight, they agreed, and with the help and guidance of our skilled meeting moderators, the FSHD Society hosted the VOPF on June 29. This was only the third time ever for the FDA to attend this type of meeting virtually, and we thus made sure that this pivotal event wasn't delayed any further. The proceedings of the meeting will be summarized in a "Voice of the Patient Report," submitted to the FDA, and used by the agency to evaluate future FSHD therapies.

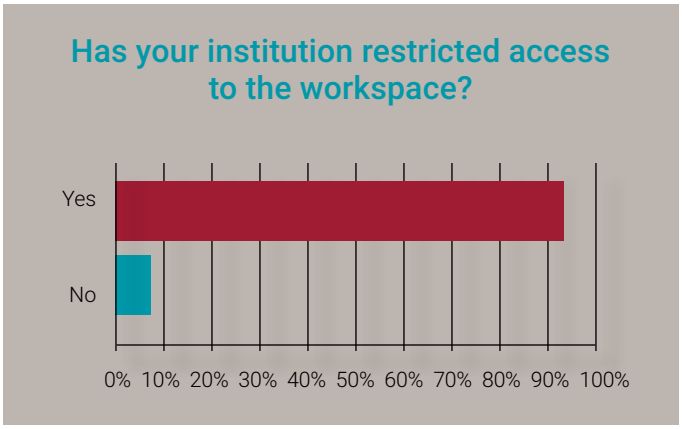
Similarly, the 2020 International Research Congress, which is a two-day event, had been scheduled for June 25-26 in Washington, DC. While other international meetings were getting canceled, the FSHD Society's IRC steering committee suggested we conduct as comprehensive

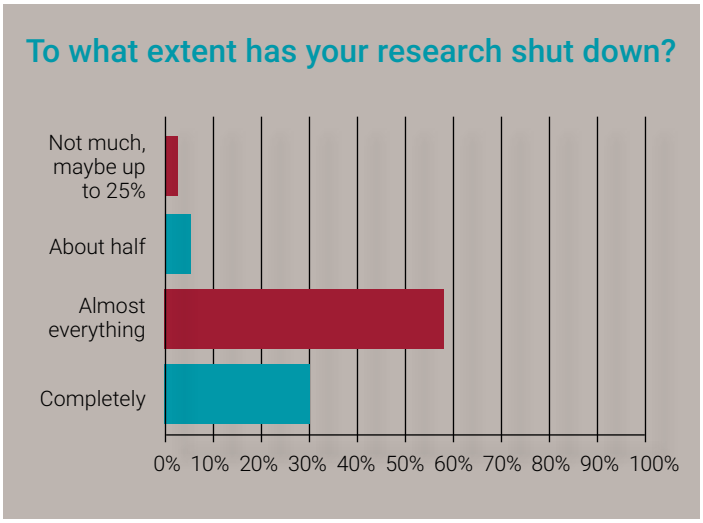
a meeting as possible to ensure that the latest research findings are shared and discussed. The 2020 virtual IRC consisted of all of the originally selected scientific presenters (more than 20!), keynote speakers, and even a virtual poster session to present preliminary data from dozens of labs.

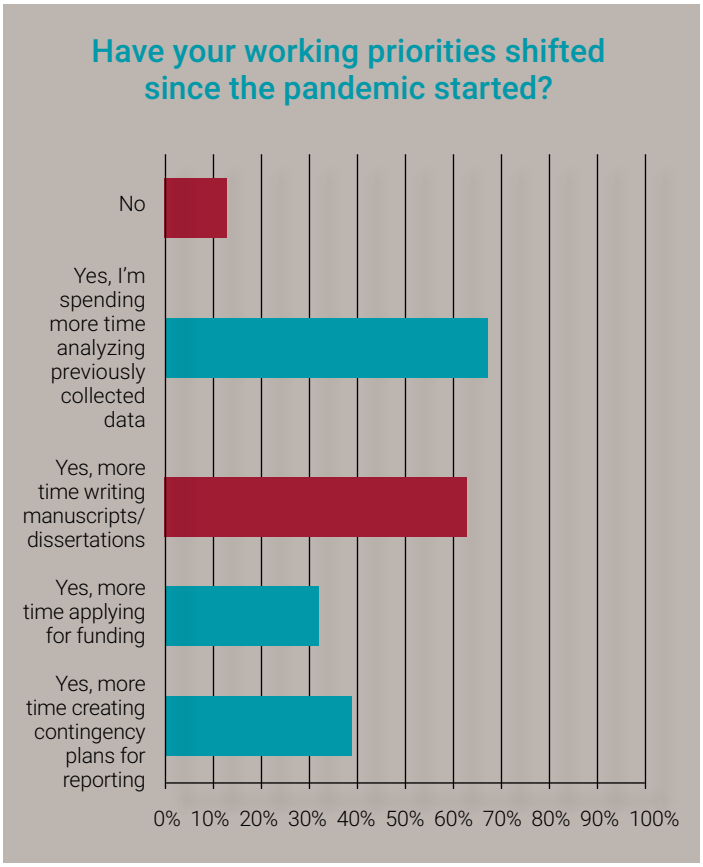
To address gaps in clinical trial readiness, a series of industry collaborative workshops had been scheduled at the rate of one meeting per quarter. While we had to postpone the first two events, we are now planning virtual workshops and will maintain the agenda, albeit in a more compressed timeline.

Lastly, to better assess how the research and clinical community have been adapting to the pandemic, the FSHD Society sent out a survey and solicited ideas and strategies to advance research. With more than 90 percent of respondents having shut down part or all of their operations, we were relieved to learn that fewer than 10 percent had experienced any furloughs. Researchers used the time away from the lab or clinic to analyze data, write papers, and draft new ideas for grant submissions. As their institutions began providing guidelines for reopening, researchers developed schedules to work in shifts to maintain safe working environments, used virtual meetings to brainstorm and review projects, and worked to accelerate research by setting up or expanding collaborations to share unpublished data. In addition, the clinical community has been incorporating telemedicine and surveys to continue serving the patient community.

One of the main points of concern shared by most researchers was whether government funding would be diverted to COVID-19 studies. No one knows exactly how the pandemic will impact future funding. However, given the Society's exclusive focus on FSHD, your generous support will continue to fund relevant research and activities to advance therapeutic developments.



# ReSolve in the face of COVID-19

*Forced to adapt, researchers have found some silver linings*

**BY JEFFREY STATLAND, MD, AND RABI TAWIL, MD**

Research has not stopped because of COVID-19. But, like all of us, it has had to adapt. The safety of our volunteers comes first.

Every study requires the doctors and research staff involved to weigh the risk of an in-person visit against the overall benefit of the study. Visits for interventional or clinical trials testing drugs (such as the ReDUX4 study) have not been canceled at most institutions. However, for observational studies such as the ReSolve study, visits have been canceled until the local authorities loosen the current restrictions based on the number of COVID-19 cases in that locale.

The ReSolve study is the largest observational study undertaken in FSHD; it has recruited 220 individuals in the US and Europe for the purpose of gleaning essential data for optimally designing future drug trials. A study like ReSolve can cost millions of dollars, and it took a decade to bring all the people together to run the study. We expected the closure to go into June, so we anticipated that many study visits would not be completed during that time. So how did we adapt?

For visits scheduled during the COVID-19 closures, we sent study volunteers the questionnaires they would have completed during the missed visit. A research coordinator reached out to the patients to help them with the process.

When each institution, with guidance from its local and state government, is ready to receive research patients, the scheduled visits will resume. To facilitate this, we have expanded the original study time window. We are asking study volunteers who are still within a three-month window around the original scheduled visit to come in once the centers reopen. For those beyond that time, we will cancel that particular visit and schedule the next expected visit. The local research coordinator will be in touch to help explain the process.

It is important to know that at all institutions, strict safety precautions will be in place. Every scheduled patient will be called ahead of time to make sure they are not ill. Their temperature will be taken when they present to the research center. They will be asked to wear a mask, and all staff will have the required personal protective equipment such as masks and gloves. We will be asking study participants to come alone, if possible, to help maintain social distancing.

Our volunteers' participation really means a lot to us, and we will do our utmost to keep everyone safe. Thanks to all who have already participated or are currently participating!

During the COVID-19 pandemic, we are also considering other ways of adapting, should the closure persist for many months. Advances in technology have made it possible to consider research visits that could occur completely at home with the use of telehealth, mobile devices, or remote sensors. Video conferencing apps allow us to assess volunteers in the home setting. Mobile devices can be used not only for questionnaires, but also to measure how people move. We have a new clinic-based study we call MOVE FSHD, which should be starting over the next six months, where we are allowing remote enrollment and piloting remote assessments. Expect to hear more about it!

Sometimes it takes a crisis to drive innovation. Two things we think have been accelerated during COVID-19 have been the development of remote assessments for research and telehealth visits. Telemedicine uses two-way video technology so doctors can examine patients in their home. We spent years trying to get telemedicine set up, but there were many institutional barriers. Now, almost overnight, we're doing it – and we hope to continue to offer telehealth once the local pandemic restrictions end. (See article on page 14.)

*Sometimes it takes a crisis to drive innovation. Two things we think have been accelerated during COVID-19 have been the development of remote assessments for research and telehealth visits.*

# You are the future for FSHD

*Help us grow our network*

**BY JUNE KINOSHITA, FSHD SOCIETY**

The FSHD community has an urgent challenge and a powerful opportunity to help support research studies and upcoming clinical trials. Currently, there are nearly a dozen very promising drug candidates. One is already being tested in patients. Others are in earlier stages. We are more hopeful than ever before that a cure is in our sight.

Past experience has shown that for each person who meets the criteria for inclusion in a study, 10 people with FSHD must be screened. The larger the number of potential participants with FSHD who are in our network, the faster and better these trials can find the volunteers they need.

In the US alone, we estimate there are 20,000 to 50,000 people with FSHD, yet we have only 4,000 such individuals in our database. How do we identify more potential participants for these critically important studies? While participation is entirely voluntary, if we don't know whom to contact, we can't share vitally important information with patients and families.

How do we identify these individuals to grow our network? This is where you can be invaluable.

First, we know many families have a "point person" who receives our emails. The others count on the point person to pass along useful information. This is not a fail-safe method. Also, our clinical trial alerts are sent only to people in the geographic location of the trial.

## What can you do to help?

First, please talk to family members and friends who have FSHD, and encourage those who are not yet in our network to join us.

Second, talk to the medical professionals who provide care to you or your family members about the FSHD Society. We hope these professionals – doctors, physical therapists, occupational therapists, orthotic makers, etc. – know of other individuals with FSHD, and can inform them about the Society and urge them to join our network. We have created a *one-page flyer* which you can download, print out, and give to your providers.

The more people we connect with, educate, and activate, the more powerful our community will be. Together we will move forward with urgent speed toward a cure.

**Editor's note:**
We thank Madeline Weinstein for her invaluable advice and input.

# What a difference two years have made!

*2020 update on our chapter program*

**BY BETH JOHNSTON, CHIEF COMMUNITY ENGAGEMENT OFFICER, FSHD SOCIETY**

It was during our 2018 FSHD Connect conference in Las Vegas that we announced the official launch of the FSHD Society's nationwide chapter program. We had three very important goals:

- Provide patients and their families with education and empowerment.
- Build our FSHD communities and connectivity to one another.
- Help fundraise critical dollars to support our mission.

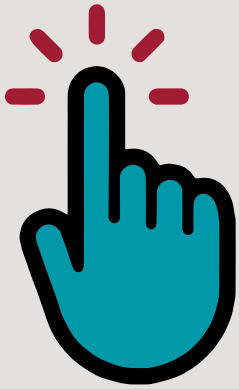
What has happened during these past two years has been nothing short of spectacular. We have launched 28 official chapters across the country! Because we at the national office cannot be everywhere, our chapters provide families with local "hands and hearts" so that no one has to face life with FSHD alone. Our chapters provide families with the education to be their own best caregivers.

Our chapters have also provided the driving force behind our signature Walk & Roll to Cure FSHD fundraisers. In 2018, they hosted five inaugural Walk & Rolls, raising more than $239,000. In 2019, we grew to 11 events, raising more than $420,000. It's now 2020, and our chapter leaders are planning 24 virtual events from coast to coast!

**Nearly 50 chapter directors and Walk & Roll leaders gathered for our 2020 volunteer leadership summit in Chicago.**

These fundraisers provide critically important dollars for our mission, but they do so much more. They provide a sense of community and purpose for our families. They allow everyone to participate in the solution, and to know that their activism has a direct impact on helping us achieve our shared goals. And they raise wider awareness of FSHD by equipping every participant with the opportunity to share their story with their families, friends, and communities.

With deeper engagement from our community, we are becoming the "army of activists" that is accelerating research toward our goal of having treatments by 2025.



## Get involved with our chapters!

Listen to some of our chapter directors and Walk & Roll leaders on what their engagement has done for them and their communities. **CLICK HERE.**

To see the list of local chapters and get to know the chapter directors **CLICK HERE.**

Don't see one in your area? Interested in starting one? **CLICK HERE.**

To see upcoming Walk & Rolls **CLICK HERE.**

Interested in hosting a Walk & Roll or other fundraising event? **CLICK HERE.**

To see our entire events calendar **CLICK HERE.**

# Together, we Walk & Roll to Cure FSHD!

*We can come together and build community, even while remaining socially distant*

**BY LEIGH REYNOLDS, FSHD SOCIETY**

When the Walk & Roll to Cure FSHD was launched in the fall of 2018, a $105,000 fundraising goal felt ambitious. One year later, we welcomed thousands of participants and raised nearly half a million dollars. Amid the global pandemic, however, sequester-at-home restrictions and public safety guidelines threatened to derail our progress.

But our remarkable volunteer leaders from across the country were not to be deterred. Collectively, we decided to transition our Walk & Roll to Cure FSHD into a virtual event that will take place nationwide on September 12.

Anyone from anywhere can register as a virtual team or individual participant. Registration gives you access to a free participant web page and state-of-the-art fundraising center. From there, you can share your story on Facebook, via email, and by text, and invite others to invest in moving us closer to a cure.

The National Virtual Walk & Roll to Cure FSHD is first and foremost a fundraiser to support the mission of the FSHD Society, but it is also about so much more:

- **Building community –** On walk day, participants from coast to coast will share on social media, call in to the live Walk & Roll radio show podcast, and join us in a collective outcry for awareness, treatments, and a cure. We will send a clear message that no one need travel this road alone.
- **Empowering patients/families –** We collectively participate in a one-day event, but we are forever changed through our connection, inspiration, and determination to fund progress toward treatments and a cure.
- **Telling the story –** Through fundraising, every participant is challenged to tell their story – how FSHD has impacted their family, how the FSHD Society has helped, and their hopes for the future. The Walk & Roll opens the door to that conversation, and leaves others more informed and aware about FSHD and its impact.
- **Building programs –** The Walk & Roll engages a wide cross-section of people with unique talents and interests that can be plugged into other chapter events.
- **Tremendous fundraising potential –** Thousands of individuals, each raising a little, add up to a lot. The funds raised through this event will help fund a cure.

## Join this nationwide effort! Visit *fshdsociety.org/walkroll* to:

- Find your local chapter's Virtual Walk & Roll and register to participate.
- No chapter near you? Join the National Virtual Walk & Roll.
- Support a walker/roller with a gift toward their fundraising efforts.
- Share information about the Walk & Roll on your social media accounts. *#NationalFSHDWalkRoll #CureFSHD*
- Tune in and share our National Walk & Roll radio show being broadcast live on Facebook *@FSHDSociety* on September 12.



**Karen Friend shows off her T-shirt.**

**At the 2019 Pacific Northwest Walk & Roll, from left to right: Nicole Bosch, Laura Jolly, Cindy Farmer, Susan Larson, Carlos Romero, Bev Branson, and Nancy Payton.**

# Genetic testing for FSHD –

BY JUNE KINOSHITA, FSHD SOCIETY

There are exciting new developments in FSHD genetic testing. A new genetic test for FSHD is now being offered by PerkinElmer Genomics, a global company known for scientific and medical testing services. Other companies are likely to follow suit. This is the first major innovation in FSHD genetic testing in nearly three decades to become available for your doctor to order.

This development has a number of advantages:

- The new test for FSHD Type 1 is expected to be two to three times less expensive than current alternatives.
- Separately, the company offers a comprehensive test for 132 other neuromuscular conditions including FSHD Type 2.
- The test includes genomics support to your physician to explain what the test means for you and your family.

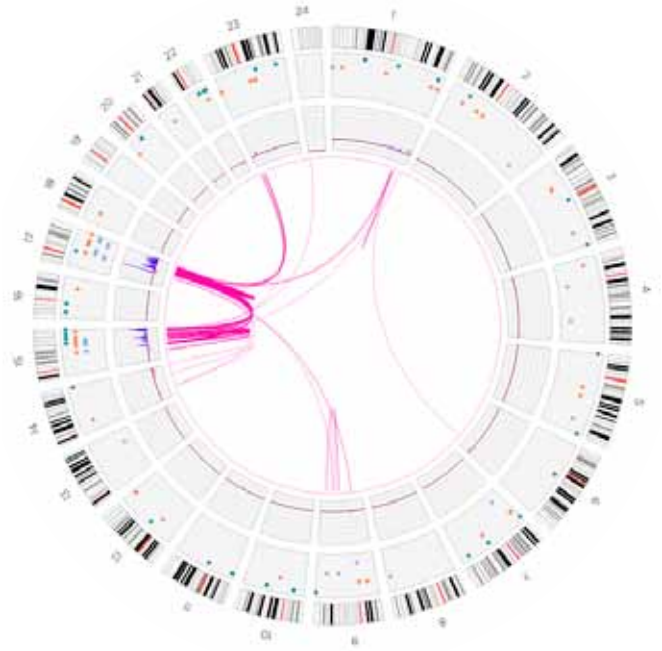
## Why seek a genetic test?

Usually, a doctor will diagnose facioscapulohumeral muscular dystrophy (FSHD) based on taking a family history and examining the patient for symptoms. For many doctors, the mindset was that "there's no treatment or cure," so the added expense of a genetic test was not warranted.

Today, a doctor's diagnosis is still perfectly valid, but there are some compelling reasons to also have a genetic test:

- The genetic test is definitive, especially if you have unusual symptoms that may cast some doubt on your diagnosis.
- You can avoid expensive and painful procedures (such as muscle biopsy and electromyography), as these are useful only for ruling out other conditions and do not confirm whether you have FSHD.
- Knowing your diagnosis for certain may help you plan for the future.
- You and your family can make informed decisions about who else in the family should get tested, how the diagnosis affects family planning, and so forth.
- Some clinical studies or trials require that volunteers have their FSHD diagnosis confirmed by a genetic test.
- The FDA could limit access to some future treatments to only people who have had a genetic test.

For a more in-depth understanding of whether you may wish to seek genetic testing for you and/or your family, please read our FAQs about genetic testing for FSHD by genetic counselor Julie Cohen, MSc. She has counseled hundreds of families at the Kennedy Krieger Institute, one of the world's leading research centers for genetic muscle diseases.



**Chromosome mapping image courtesy of Bionano Genomics.**

## How is the new test different?

The cause of FSHD lies in our genome, the DNA molecules that are bundled up into 23 chromosomes residing in the nucleus of the cells in our bodies. Our genomes contain the "genetic code" – the molecular compendium of instructions that makes us human and unique.

FSHD Type 1 results from the shortening ("contraction") of a stretch of DNA near the tip of chromosome 4. The "FSHD region" on chromosome 4 consists of many units called D4Z4, which are repeated like beads on a string. Having more than 10 units is protective, but individuals who have fewer than 10 are at risk for FSHD. The reduced number of D4Z4 units results in an increased chance for the expression of a gene called *DUX4*, which is normally locked up and silent in adult cells. When *DUX4* gets expressed, it leads to damage and death of muscle cells.

For many years, the test for FSHD1 involved a tech-

# A new frontier

nology called Southern blot (named after its inventor, the English biologist Edwin Southern). This technology involved taking a person's DNA (typically extracted from blood cells), chopping the long DNA strands into smaller pieces, and separating the pieces by their size to roughly estimate the number of D4Z4 repeats. Southern blots take a lot of labor and time, and at best provide only an estimate of the number of repeats.

Now all this is changing with the new test. Using a technology called whole-genome optical mapping, developed by Bionano Genomics, scientists are able to examine very long strands of DNA, which are stretched out like pieces of thread in a super-thin tube and labeled with a fluorescent tag so that the number of D4Z4 units can be directly counted under a microscope.

## Research versus clinical tests

What should you do if you cannot get a genetic test through your doctor? There are a variety of reasons why this situation could arise:

- The doctor won't order the test.
- Insurance won't cover the cost.
- The individual doesn't want the genetic test result in their medical record.

If, based on symptoms or family history, a person qualifies to join an FSHD research study or clinical trial, they may be able to obtain a genetic test at no cost. Each research study has its own goals and protocols, so it's important to understand what kind of testing is being offered and how that information can or cannot be used.

Volunteers participating in the National Registry for FSHD at the University of Rochester, for example, can have their DNA tested in the Netherlands. (The FSHD Society and Friends of FSH Research have funded this program.) A positive test helps genetically confirm your doctor's clinical diagnosis, but because the test is not done in a certified commercial lab, it is not considered an official genetic confirmation.

Another opportunity to have genetic testing as part of a research study is through the laboratory of Peter and Takako Jones at the University of Nevada, Reno. Their test is based on the "methylation" of the D4Z4 region. "Methylation" is a molecular tag on the DNA that normally keeps genes from being expressed. In unaffected people, the D4Z4 regions are heavily tagged, but in FSHD1 or FSHD2, there are fewer tags ("hypomethylation"), which leads to an increased risk of *DUX4* becoming expressed and causing disease. This test can be done using saliva samples that are collected in a special tube and simply mailed to the Jones lab. People don't have to get their blood drawn. The test is still experimental, and the Jones lab is in the process of collecting samples from 1,000 volunteers, including individuals with FSHD, their healthy family members, and people with other neuromuscular conditions. The methylation test is viewed as highly promising by scientists, but there is not a broad consensus yet that it should serve as an FSHD diagnostic test on its own.

To date, limited access to FSHD genetic testing poses a serious problem for research. Just a few years ago, nearly half of the individuals in the national registry had not had a genetic test. Without it, the rest of their data are of limited value. The large percentage of patients who have not had genetic testing also slows down recruitment for clinical trials.

The FSHD Society has placed a high priority on having a genetic testing system that is affordable and accessible while capturing data that researchers need to better understand FSHD and develop treatments. One future scenario is to screen individuals for hypomethylation of the D4Z4 region with a fast, inexpensive test. People with low methylation consistent with FSHD would be referred for more comprehensive testing for the number of D4Z4 repeats and the presence of other mutations or genetic modifiers. Patients could agree in advance to allow their genetic and clinical data to be studied for research.

The data from such a system would yield invaluable insights for new treatments and future clinical trials. Our goal is to bring together the scientists, companies, patient advocates, and other stakeholders to make this vision a reality.

Visit our blog post, *Genetic testing for FSHD – a new frontier,* for reasons to get genetic testing, where to get tested, and for a letter to request insurance coverage of genetic testing.

# Telehealth for FSHD

BY JUNE KINOSHITA, FSHD SOCIETY

Telehealth for FSHD is an idea whose time has come. If people with FSHD are rare, doctors who understand FSHD are rarer still. Patients who don't have the time or resources to travel long distances may never have the opportunity to consult a medical professional with FSHD expertise. That is now changing. One of the silver linings from social distancing during the COVID-19 crisis is the swift and broad embrace of telehealth by the healthcare sector.

With telehealth or telemedicine, a doctor or other healthcare professional can set up a two-way video call with the patient. They can assess the patient's condition based on what they see and hear over the call and provide medical advice.

The doctors who lead the *FSHD Clinical Trial Research Network (CTRN)* have been at the forefront of this change. In our recent webinar, Dr. Jeffrey Statland, co-director of the CTRN, mentioned that he is offering telemedicine consults. In follow-up, the FSHD Society worked with Kiley Higgs at the CTRN to compile a list (see below) of the CTRN sites and others that are offering telemedicine, along with contact information for scheduling an appointment. *Visit our web page* for updates.

## List of CTRN and other sites offering telehealth

| Institution | Clinic Director | Phone | Fax | Email |
|---|---|---|---|---|
| Kennedy Krieger Institute | Kathryn Wagner, MD PhD<br>Julie S. Cohen, genetic counselor | | | *lasseth@kennedykrieger.org*<br>Make appt online Link:<br>*https://www.kennedykrieger.org/patient-care/centers-and-programs/center-for-genetic-muscle-disorders* |
| The Ohio State University | Samantha LoRusso, MD | (614) 293-4969 | (614) 293-6111 | |
| UCLA | Perry Shieh, MD PhD | (310) 794-1195 | (310) 794-7491 | |
| University of Kansas | Jeffrey Statland, MD | (913) 588-6820 | | |
| University of Massachusetts | Lawrence Hayward, MD PhD | (508) 334-2527 | | |
| University of Rochester | Rabi Tawil, MD | (585) 273-2743 | (585) 273-1255 | |
| University of Utah | Russell Butterfield, MD | (801) 585-7575 | | |
| Virginia Commonwealth University | Nicholas Johnson, MD, and Chelsea Chambers, MS LCGC | (804) 482-1260 | | *Chelsea.Chambers@vcuhealth.org* |

# Biopharmaceutical industry news

## Dyne Therapeutics ramps up its FSHD program

Dyne Therapeutics, a biotechnology company based in Waltham, Massachusetts, announced the acceleration of its programs in FSHD through the exclusive licensing of technologies to target the genetic basis of the disease, as well as the appointment of leading researcher Jeffrey Statland, MD, to its Scientific Advisory Board. Statland, an associate professor of neurology at the University of Kansas Medical Center, is the co-principal investigator for ReSolve (Clinical Trial Readiness to Solve Barriers to Drug Development in FSHD), an ongoing observational study run by the FSHD Clinical Trial Research Network (CTRN) and supported by Dyne Therapeutics.

Dyne's proprietary FORCE™ platform enables targeted delivery of oligonucleotides and other molecules to skeletal, cardiac, and smooth muscle. It has achieved promising results in mouse models of myotonic dystrophy type 1 (DM1) and Duchenne muscular dystrophy (DMD).

The company's therapeutic approach to FSHD utilizes the FORCE platform to deliver an oligonucleotide targeting the gene *DUX4*, which is the genetic basis of FSHD, inside the muscle cells of FSHD patients. After entering the muscle cells, the oligonucleotide is expected to reduce aberrant expression of the DUX4 protein and halt the loss of muscle function that characterizes FSHD. The intellectual property exclusively licensed by Dyne is based on oligonucleotide technology developed by Alexandra Belayew, PhD, and Frédérique Coppée, PhD, at the University of Mons in Belgium. FSHD Society grants helped to fund the basic research underlying this technology.

**Members of the Dyne team.**

## ReDUX4 adapts to COVID-19 challenges

The COVID-19 pandemic could not have come at a worse time for ReDUX4, the Phase 2b clinical trial of losmapimod, a drug that is being developed by Fulcrum Therapeutics as a potential treatment for FSHD. Yet the Cambridge, Massachusetts, biotech rose to the challenge, nimbly adapting its protocol to enable the trial to continue. What's more, by extending ReDUX4 by an additional 24 weeks, the trial will collect data over a longer period, which, given the slow progression of FSHD, may improve the likelihood of detecting effects of the drug.

"In the wake of COVID-19, a number of our clinical trial sites postponed their activities," said Robert J. Gould, PhD, Fulcrum's president and CEO, in the company's first-quarter report to investors. "Our team acted swiftly to minimize the health risks to patients, families, and healthcare professionals involved in our studies of losmapimod in the treatment of facioscapulohumeral dystrophy (FSHD). We amended our Phase 2b trial, ReDUX4, to extend the trial from 24 to 48 weeks; added an interim analysis for subjects who underwent their 16-week biopsy as originally planned; and added a 36-week biopsy for patients who cannot undergo their 16-week biopsy. We believe these changes will enable patients and investigators to continue participation in the trial and allow us to collect the essential data needed to evaluate the potential efficacy and safety of losmapimod for the treatment of FSHD."

# Finding a way to keep working

*Help is out there, but it takes perseverance to get it*

**BY LYNN STEVENS, BOSSIER CITY, LOUISIANA**

About 10 years ago, the progression of FSHD on my body forced me to begin evaluating whether I should continue trying to work full-time or consider what was next for me. I couldn't imagine sitting on the sidelines, as this was not in my DNA (pun intended).

But after several bad falls – one that resulted in an ambulance ride and having to get my teeth put back in and my mouth wired shut – I had to face a grim reality. Until then, I had tried to ignore what was happening, mostly because I was scared to admit that I was at this point, and I had bills to pay. I couldn't just stop working, but what were the alternatives?

> *"Many of us have or will have to face the same decision at some point in our lives. You are not alone – that is the most important thing you need to remember. You also have to find what works for you. There are help and programs out there for you, no matter where you live."*
>
> **– LYNN STEVENS**

It actually took me a long time to get to where I am today. I could have just reduced my hours or my job duties and responsibilities, but that was not me. I was raised by a Marine dad who instilled the "never give up" mentality.

Instead, I used the resources with my employer at the time, the United Way, to help me. Being connected to this large umbrella community organization, I tapped into the resources in our own lobby to find what options I may have.

Our office often referred disabled individuals to the local Rehabilitation Services office. Every state has its own version that offers vocational services free of charge to those who apply and are eligible. Every state has different eligibility requirements and funding levels. This is all I really knew about the services or programs it offered. After facing my fears of admitting that I needed help, I made the call.

I learned that, because I wanted to keep working, there were resources that I qualified for immediately, including help securing a wheelchair and funding to convert our van. They even did a home and workplace assessment, and helped me purchase items for my house and for my employer to better accommodate my ability to continue in my current position.

It wasn't all roses, though. It was a painstakingly slow process, taking more than a year for me to get the help I needed. I learned more about self-advocacy than I had in 20-plus years of having FSHD. But I did get the help I needed at the right time in my life.

About a year ago, I started facing a new battle: severe fatigue. It started to impact everything – my work, social contacts, and quality of life. Again, I was forced to face facts. It was time to reduce my hours or quit working. Unfortunately, as the chief operating officer at United Way, I didn't have the option of reducing my hours. But I really didn't want to stop working. So now what?

One of my final projects with the United Way was helping to write a collaborative grant in partnership with Goodwill Industries. Even though I was an avid shopper

at Goodwill, I never thought it would present a career opportunity that would become the next chapter in my life.

Goodwill Industries works to enhance people's dignity and quality of life by strengthening communities, eliminating barriers to opportunity, and helping individuals reach their full potential through the power of work. The Goodwill network represents 157 community-based organizations in the US and Canada, with a presence in 12 other countries.

Each of the independent, local Goodwill organizations designs its own programs and services to employees, and helps people find work where they live. All of them offer programs and services for individuals with disabilities. And even though the United Way had funded them for years, this is something I never knew.

Since October of last year, I have had the pleasure of working as the director of Workforce Development – a role I have loved since day one. Because the organization is about serving those with disabilities and disadvantages, I have never feared my disability or how it affects my employment. Our CEO is very understanding and has worked hard to make sure everything I need is available. The best things about this role are the satisfaction I get every day helping others achieve their employment goals – whatever that looks like for them – and helping to educate employers on how to work with the disabled and advocate for community-wide changes.

Many of us have or will have to face the same decision at some point in our lives. You are not alone – that is the most important thing you need to remember. You also have to find what works for you. There are help and programs out there for you, no matter where you live.

**Editor's note:**
Lynn Stevens is the director of Workforce Development for Goodwill Industries of North Louisiana and has been serving the nonprofit sector for more than 20 years. She has used her experience and leadership skills to help guide and direct many types of local, regional, and national organizations.

## Resources to help find a job

**American Job Center Finder**
*careeronestop.org/localhelp/americanjobcenters/find-american-job-centers.aspx*

**State Vocational Rehabilitation Programs**
*parac.org/svrp.html*

**Directory of Centers for Independent Living and Associations**
*ilru.org/projects/cil-net/cil-center-and-association-directory*

**State Labor Offices**
*dol.gov/agencies/whd/state/contacts*

**Office of Disability Employment Policy: State Liaisons**
*dol.gov/odep/contact/state.htm*

**AbilityOne Network: Employment Opportunities**
*abilityone.gov/abilityone_network/employment.html*

**Goodwill: Jobs and Training**
*goodwill.org/jobs-training*

# "Eddie-mechanics" was my secret to success

**BY ED BAROCAS, MONTCLAIR, NEW JERSEY**

In spring of 2019, I received two awards for my legal work: the New Jersey Law Journal's Attorney of the Year Award, and the American Civil Liberties Union's Roger Baldwin Award for a lifetime of defending civil liberties. The ceremonies were beautiful but bittersweet. I had announced my retirement months earlier as the exhaustion, discomfort, and worsening deterioration of my muscles caused by FSHD as well as my related digestive issues had simply become too great.

The ACLU event essentially served as my retirement party. During my career I never felt completely comfortable with or even deserving of high levels of praise. But that night, colleagues from my 25-year career lauded me. I felt like Jimmy Stewart in *It's a Wonderful Life* being given the gift of realizing that your life has had meaning and a positive impact on others.

Some people commented that they were all the more impressed because I accomplished what I did "despite my disability." I always recoil a bit when I hear that phrase. Clearly, my disability created numerous obstacles for me. But living with FSH muscular dystrophy helped shape who I am. I wondered, Would I have become the same person – and would I have been as successful in my career – were it not for my disability?

As a child, I knew there was something different about me. There were not many noticeable physical symptoms of FSHD except that I shared with a number of family members the same facial features and inability to move my upper lip. I always had a close family, a strong personality, and many close friends, so knowing I was somehow different did not devolve into feeling like an outcast. The understanding of being different was instead directed outward, as empathy for others who were different from "the norm."

> *"Would I have become the same person – and would I have been as successful in my career – were it not for my disability?"*
>
> — ED BAROCAS

Because I had the security of family and friends at a young age, I felt empowered to speak out for other "outside-of-the-box-ers" who were bullied or excluded and were afraid or unable to speak up for themselves. From grade school and beyond, I reveled in taking up the cause of the underdogs. I never felt such a sense of strength as when I would stand up for someone who was being wronged, especially when I succeeded in setting things right.

That sense of strength and control was quite the opposite of my physical being. I was always one of the slowest and weakest boys. During gym class, others might joke about it, not recognizing yet that it was due to a disability. (And I didn't fully understand it either; nor was I comfortable talking about it until my early teens.) But I had excellent eye-hand coordination, so I could hold my own when it came to certain sports, especially tennis. By the time I turned 12, my muscular dystrophy had started to really hit my arms and legs, and I continued to become slower and weaker. In order to win on the tennis court, I had to learn to adapt. I had to engage in what my friends call "Eddie-mechanics."

Eddie-mechanics applied to almost everything physical (and, as it turned out, beyond the physical). I often could not do even the most mundane of tasks the normal way. I had to learn to step back, look at the big picture, consider a new approach, and find my own way. When it came to tennis, it meant not only crafting a good serve without lifting my arm over my head, but also using different spins and angles instead of power or speed, finding new tactics, and making it a mental game rather than a physical one. I would need to

Photo by Carmen Natale, courtesy of the *New Jersey Law Journal*.

quickly identify the flaws in my opponent's game and figure out how to exploit them. I had to improvise on the fly if I wanted to win. And I always wanted to win, to show that nothing could stop me. I lost some but won more than my fair share of games, leaving my opponents angry or confused as to how they lost to me.

My need to prove that "nothing could stop me" was, however, at times counterproductive and nothing more than stubborn pride. Indeed, up until my early 30s, I was not completely comfortable with having FSHD. I only saw how it impacted me negatively, and still viewed any success as "overcoming" my disability. And my drive to do so could blind me. Especially when it came to going into a wheelchair. By 30, I walked with quite a funky limp, my back was arched, and I relied on a walking stick. I could not stand for more than 10 minutes. I had trouble getting up from a seated position. A few times in a year my back gave out, and I would have to wait for someone to help reposition me. I never went out in the winter except to work, and the short walk from my car to the front door of my apartment was fraught with anxiety.

Then, in my early 30s, while driving on a gorgeous sunny day, I found a hidden garden near my home. There were flowers, trees, rolling hills, and the smell of a cool breeze. But the garden's grounds were uneven, and the fear of falling grew with every step. I could only survey the beauty from afar for a few minutes, and then turned back to my car. Once home, I sat on my balcony as the sun set, called an old friend, and relayed what had happened. "Don't take this the wrong way, Ed," he said, "but wouldn't going into a wheelchair be liber – " He had not finished saying that word when a wave of clarity – that "A-ha!" moment – occurred. In a flash, I understood that going into a chair would not be confining; it would be liberating. It had been my own stubbornness that had confined me.

Well before I recognized the unique influence my disability had on developing my psyche and skills, my career path was clear. I entered law school knowing that I wanted to spend my life defending the rights of people who were viewed as different, and ensure every person is treated fairly and justly.

After law school, I was hired by the public advocate's office representing mental health patients who, having been deemed mentally ill and dangerous, were committed to psychiatric hospitals. It was then that I discovered an unexpected benefit from having my disability. Although the job was to represent the patients, they often did not trust us, and viewed us as cogs in the system that was keeping them in a hospital against their will. But given my obvious disability, the patients did not see me that way. As one patient said: "Do you know why we trust you? You are one of us." They saw me as a fellow outsider. That perhaps allowed them to open up to me, and to help me help them.

After a few years in that unit, I was selected by the public advocate's office to represent sex offenders in Megan's Law community notification hearings. The head of the unit and I ended up bringing numerous constitutional challenges to the law. With little experience in such complex work, we were proceeding by the seat of our pants. Having to solve problems or overcome roadblocks on the fly had become second nature for me. My superiors became comfortable giving me more and more responsibilities, and in just a few years I gained invaluable experience, arguing cases before the New Jersey Supreme Court and United States Court of Appeals.

After five years in that position, I was hired for my dream job: legal director of the American Civil Liberties Union of New Jersey. That position fulfilled my passion to help the underdogs and the voiceless. But it also was the position that, more than any other, best utilized not only the skills I learned in law school, but the skills I developed from living my particular life.

Working on constitutional law issues requires the ability to approach things logically and creatively at the same time. The work of the ACLU is unique. It is not to merely see that existing precedent is enforced, but to advance the law. One must step back, put aside status quo thinking, and find an approach that is distinctive and new but that can be so strongly defended that the courts will accept it. Just as in my tennis matches, it requires crafting new spins (on prior court decisions) and finding the

***Continued on page 21...***

## Sequester Camp webinars

Available on our *website, Facebook video,* and *YouTube channel.*

**March 19 –** We are in this together
**March 26 –** How to connect. How to help
**April 2 –** Life hacks in the age of coronavirus
**April 9 –** COVID-19, FSHD, and the impact on research
**April 16 –** Caring for ourselves during difficult times
**April 23 –** Patient-reported symptoms in FSHD
**April 30 –** Staying active while at home
**May 7 –** Exploring the national registry for FSHD with artificial intelligence
**May 14 –** Getting more sleep during restless times
**May 21 –** Fulcrum's preparatory studies for clinical trials
**May 28 –** A new genetic test for FSHD
**June 4 –** Adapting your home for safety
**June 11 –** Stem cell research in FSHD

FSHD SOCIETY SEQUESTER CAMP

## Connecting our community through this crisis

### Fitness with Mitch and Bill

Our fitness workout webinars with Bill Herzberg *(billherzberg@gmail.com)* and his trainer Mitch Wade *(mwadefit@gmail.com;* on Instagram *@mitchwadepdx)* were quite the hit at Sequester Camp. Held on Mondays, the webinars discussed the progress Bill has made in addressing his FSHD muscle weakness by working out with Mitch. All of their webinars are available on our *Facebook video* and *YouTube channels.*


**Mitch Wade and Bill Herzberg at Mitch's Portland, Oregon, gym, Ramp Fitness.**

### How can you help? Make a donation!

The FSHD Society is almost entirely funded by donations from individuals and families like yours who have a personal stake in advancing research. Together, we are accelerating the development of treatments and a cure.

### Double your donation

By simply completing the matching gift form from your employer, you may be able to double, or even triple, the impact of your gift! Find out if your company will match your gift **HERE.**

### Get Social!

Our online communities are great sources of news, advice, and social support. Follow us on *Facebook, Twitter,* and *Instagram*.

### Volunteer for research

One of the most powerful things people with FSHD can do is to volunteer for research studies. Your participation is vital to advancing health care, diagnosis, and treatments. Visit our *Clinical Trials page* for more information.

### Not getting our emails?

Don't miss out on important news. *Sign up here.* If you are certain you are on our email list, please check your spam or junk folder.

## Commit to the future

Take your support to the next level by including the FSHD Society in your will. Your bequest sustains our work for future generations. Questions? Please contact Heidi Roy at *heidi.roy@fshdsociety.org* or (781) 301-8530.

Always check with your financial advisor when making a change in your will or estate plans.

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## "Eddie-mechanics" was my secret to success

best angles to win your points.

I was conditioned to notice every crack or imbalance that could upend a case, to see every potential roadblock, and to know what logical order one should take in crafting the best course of action. My greatest talent was in editing others' work – to make a bad brief good and a good brief great – and preparing others for oral argument. In the last two decades of my legal career, we were able to alter the status quo to one that was at least slightly more fair and just.

So would my life have followed a similar path if not for my disability? Would I have developed such a strong sense of empathy? Would I have wanted to focus my career on advocating for those who were different, for the voiceless, for the underdogs? Would I have been as creative? Would I have been able to see the big picture, to find unique approaches when others might not? In short, would I be close to the same person I have been? Luckily, I've been happy with who I am and what I have accomplished. So it is perfectly okay that we will never know.

# Upcoming

**For updates and registration, visit *fshdsociety.org/fshd-events-calendar/*.**

### WEBINARS

**July 16, 1–2 p.m. ET. – *Well-being***
Ora Prilleltensky, PhD

**August 20, 1–2 p.m. ET. – *Scapular stabilization surgery***
Anthony Romero, MD

### CHAPTER MEETINGS

***Connecticut Connections***
Open to all New Englanders (and beyond), this chapter meets via web conference on the first Thursday of each month (except in summer), 7-8:30 p.m. EST.

***Idaho Chapter Launch***
July 18, 12-2 p.m. MT.

***Western Washington FSH Community***
Meets via web conference on the fourth Saturday of each month (except in December), 10 a.m. PST.

### RADIO

***FSHD Society Radio***
First Wednesday of every month at 9 p.m. ET | 8 p.m. CT | 7 p.m. MT | 6 p.m. PT via Facebook Live. Podcasts are recorded and available in the video section of the FSHD Society Facebook page.

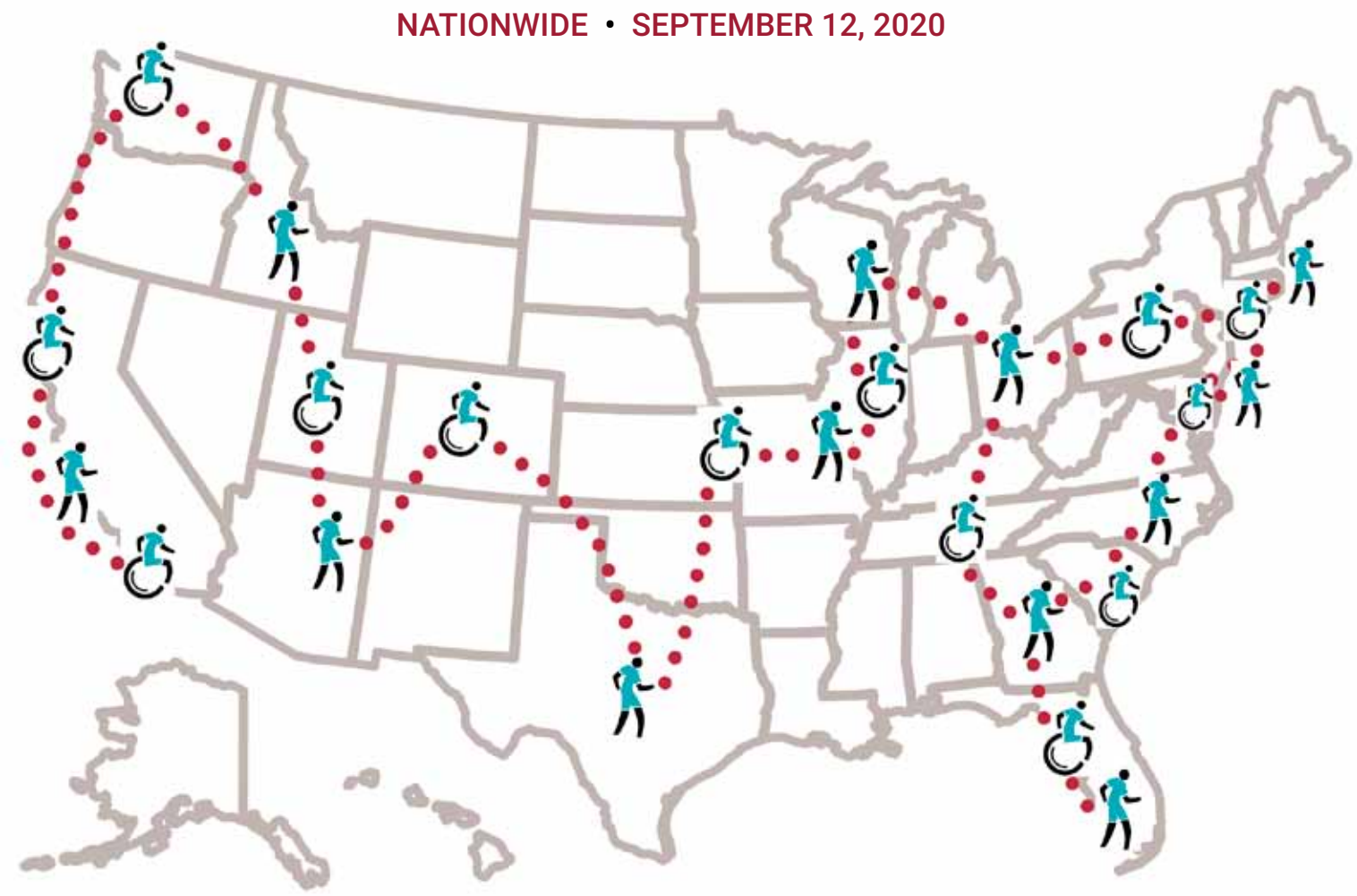

### FUNDRAISERS


**September 12**
All *virtual Walk & Roll to Cure FSHD* events by the communities listed below. Don't see one in your area? You can join the National Virtual Walk & Roll.

- Arizona
- Atlanta
- Bay Area
- Central Texas
- Chicagoland
- Colorado
- Columbus
- Connecticut
- East Tennessee
- Greater Philadelphia
- Idaho
- Kansas City
- Los Angeles
- Mid-Atlantic
- New England
- North Carolina
- Pacific Northwest
- South Carolina
- Southwest Florida
- St. Louis
- Tampa
- Utah
- Wisconsin

NATIONAL VIRTUAL WALK & ROLL TO CURE FSHD

## NATIONWIDE • SEPTEMBER 12, 2020



This year, dozens of local events have united under the umbrella of our *National Virtual Walk & Roll* on September 12. Anyone from anywhere can register as a virtual team or individual participant. Registration gives you access to a free participant web page and state-of-the-art fundraising center. From there, you can share your story on Facebook, via email, and by text, and invite others to invest in moving us closer to a cure. Join this nationwide effort! Visit *fshdsociety.org/walkroll* to:

- Find your local chapter's Virtual Walk & Roll and register to participate.
- No chapter near you? Join the National Virtual Walk & Roll.
- Support a walker/roller with a gift toward their fundraising efforts.
- Share information about the Walk & Roll on your social media accounts. ***#NationalFSHDWalkRoll #CureFSHD***

Tune in and share our National Walk & Roll radio show being broadcast live on **Facebook** ***@FSHDSociety*** on September 12.