

# Dawn of a new era

The age of clinical trials has arrived

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Look for us on the Internet at [fsbdsociety.org](http://fsbdsociety.org).

We thank the FSHD Society staff for their editorial assistance.

# The dawn of a new era

*Clinical trials in FSHD provide unique opportunities and renewed hope*



*“Every great dream begins with a dreamer. Always remember, you have within you the strength, the patience, and the passion to reach for the stars and change the world.”*

– HARRIET TUBMAN

The past three years at the FSHD Society could be characterized in the following (somewhat oversimplified) manner:

- 2020 was a year of crisis
- 2021 was a year of adaptation
- 2022 was a year of transition



Mark A. Stone

This year will be the dawn of a new era in drug development for FSHD. With more than 20 companies actively working on treatments, the FSHD community has moved into a new phase – with multiple drug trials set to launch in the coming months.

There will be challenges to overcome, along with opportunities and obligations to continue working to “de-risk” trials, reduce the barriers to trial recruitment, and ensure that, once a treatment is approved, families will have access to it. This will be a time to embrace new initiatives and methods that will enhance and continue accelerating clinical research on all levels.

It’s said that “knowledge is power,” but it is the application of knowledge that reshapes the future. As you read through these pages, you will hopefully gain a greater

understanding of the clinical trial process (p. 10), how your voice can be magnified, and your impact multiplied (p. 7). We also take a peek beyond the horizon to new hope, as research in regenerative medicine seeks not only to slow the disease but to restore muscles (p. 13).

We are at a defining moment – a new era – in the history of FSHD. Five years ago, we encountered a similar crossroad. Through sacrifice and bold leadership, we launched programs that have brought us to this point in the clinical development process in FSHD. We will require the same sacrifice and leadership to confront the challenges at hand, overcome and eliminate the obstacles, and achieve our collective mission. We did it before, and I am confident we can do it again.

Embracing the new era,

Mark Stone

President and CEO  
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# The challenges of the new era

*What can you do to help?*

BY JUNE KINOSHITA, FSHD SOCIETY

**W**hen we say that we are embarking on a new era in clinical trials, what do we mean? After all, we have seen previous FSHD trials, including those by Wyeth, aTyr, Acceleron, and Fulcrum's ReDUX4. What makes the current era different is that, for the first time, we have multiple trials running in parallel, including one in Phase 3, which is the furthest stage of development any therapeutic trial in FSHD has ever gotten!

Amongst the ongoing trials, REACH (Fulcrum's Phase 3 trial) will enroll 230 individuals with FSHD this year, while the MOVE and MOVE+ natural history studies for FSHD are seeking 450 volunteers. In addition to these ongoing studies, both Roche Pharmaceuticals and Avidity Biosciences are planning to launch their own Phase 1/2 studies in 2023, each requiring about 50 participants.

Finding more than 700 people to volunteer may not seem like a tall order until you consider this sobering statistic: Across all 20+ Clinical Trial Research Network (CTRN) sites in the US, Canada, UK, and Europe, there are roughly 2,500 patients. But only about 10 percent – or 250 – of these patients might meet the inclusion criteria for a clinical trial. As a community, we will have to work hard to fill the need for just this year. This is our immediate challenge.

If a clinical trial fails to show efficacy, that's a huge disappointment, but it can provide valuable insights about whether the selected outcomes were sensitive or whether the trial duration was sufficient. This type of information can further encourage other companies to improve their trial designs to increase their chances of showing an effective outcome.

But if a trial cannot be completed at all because it was unable to recruit enough volunteers, that is the worst kind of failure, because it discourages other

companies from taking the risk to develop a drug.

This is the crux of the problem – not having enough engaged individuals in the FSHD community to quickly fill up the clinical trials that are coming. If we do not solve it, the longer-term consequences could be dire.

## Your efforts make a difference

**You will help grow the size of our active community.** The FSHD Society reaches approximately 5,000 of you who have FSHD. Of this number, about 700 regularly engage when we invite you to participate in studies and meetings. You are the lifeblood of the community!

But another 4,300 may skim this issue of the *Advocate* or only briefly scan our emails. We understand that people have a lot of other things going on in their lives. They don't want everything to be about FSHD.

But a lot is happening now that can impact your future. *In a rare condition like FSHD, you cannot assume someone else will step up to the plate.*

**Your participation in research may enable more people to be included in clinical trials.** It seems like a paradox to worry about filling clinical trials when so many who wish to volunteer are turned away. Why is this?

Clinical trials aim to prove whether patients' strength or function stabilizes or improves when they are on the drug, so the trial must eliminate factors that can make the


data difficult to interpret. These factors include patients' age, ability to perform the required tests, ability to make trips to the trial site, which specific muscles are affected, and so on. By the time potential volunteers are screened for a trial, perhaps 9 out of 10 will not meet the criteria.

Can anything be done to broaden the criteria, for example, to include younger or older patients, or wheelchair users? Possibly. If solid data existed on the natural progression of FSHD in large numbers of older adults, children, and non-ambulatory individuals, then designing trials that include these people should be possible.

Unfortunately, there aren't enough data yet, which is why the MOVE and MOVE+ natural history studies are open to all comers. But to date, very few older people, children, and non-ambulatory individuals have enrolled. Unless many more underrepresented patients enroll in natural history studies, future clinical trials will likely continue to exclude these groups.

**You can help find the “missing” people with FSHD.** There are an estimated 50,000 to 100,000 people with FSHD in the US, EU, and UK. Advocacy groups and clinicians are connected with just a fraction of them.

We can only speculate about where the rest of them are. Perhaps half of the people who have FSHD have not been diagnosed or have been misdiagnosed with another condition. Others have received a diagnosis and were told “there’s nothing to be done,” so they never returned to the doctor. They went on living as best they could. They never connected with advocacy groups or heard about progress toward better treatments.

Some of these may be people in your own family. Or they may be your friends through social media groups. They may have symptoms but no diagnosis. Or be newly diagnosed. Or were diagnosed decades ago but never engaged with the FSHD community. Talk to them and let them know how important their participation would be to advancing treatments for FSHD. You are the most powerful advocate when it comes to connecting our community with the larger universe of people with FSHD. 

## What can you do?

- **See your neurologist.** Ask your doctor to look up FSHD trials on *clinicaltrials.gov* and discuss whether you are a candidate for a clinical trial. It's also a good idea to see your doctor to stay proactive about your health.
- **Get seen at a Clinical Trial Research Network site.** You'll get high-quality care and meet the researchers who are running trials. They can guide you if you want to enroll in a trial or other studies.
- **Join a study!** There will be three clinical trials this year. Learn about them and ask yourself if you'd like to be involved. If a trial is not right for you, sign up for the MOVE or MOVE+ studies, which are vital for future drug development efforts. See our article on page 10 on how to learn about trials and studies.
- **Talk to others with FSHD.** Most people with FSHD are on the sidelines. Talk to them about why it's so important to participate in research.
- **Read our emails.** This is the main way we alert you about new trials as well as advances in research and our wonderful community events..
- **Visit the FSHD Society website for updates.** Our blog will keep you up-to-date on the latest news.
- **Make sure we have your complete and current information.** That way you will never miss an important



announcement. Use the QR code or join at [FSHDSociety.org/FSHDRegistry](https://FSHDSociety.org/FSHDRegistry).

# Finding your rope team

*When the going gets tough, your team will hold you up*

BY BETH JOHNSTON & LEIGH REYNOLDS, FSHD SOCIETY



**A**rmy Command Sgt. Maj. Gretchen Evans earned the highest rank an enlisted soldier can achieve. She was responsible for all security and personnel in Afghanistan, overseeing more than 30,000 ground troops, until enemy fire left her with a brain injury, PTSD, and total hearing loss, ending her 27-year career in the military. CSM Evans had lost her purpose and found herself in deep despair.

Eventually, Evans connected with other survivors and formed Team Unbroken, made up of mixed-ability athletes. She persuaded a skeptical TV producer to let them compete in the World's Toughest Race: Eco-Challenge Fiji. Their feats were filmed for a documentary that is streaming on Amazon Prime Video.

We had the honor of meeting CSM Evans when she gave the keynote at our 5th Annual Volunteer Leadership Summit on a February morning at the Chicago Airport Hilton. It was frosty outside, but inside our meeting room, everyone was on fire. Sixty of our volunteer leaders from across the US and Canada gathered in person and on Zoom for two days of intensive learning, heartfelt camaraderie, open collaboration, and a little bit of fun.

Evans shared how she fought her way back to a meaningful life when she found her “rope team” who, like members of a rock-climbing team, commit to standing firm and holding you up when you need it most. She urged our own mixed-ability fighters to “find your rope team.”

*“With this kind of leadership, I feel I am part of a raging fire! After listening to what you had to say, I feel like there is nothing I can't do...”*

— HELOISE HOFFMANN

“If someone slips, the rest of us pick you up,” she explained. “There’s safety and accountability in that. You have to be connected to other people in your life who are committed to you.”

Bringing the FSHD community together, literally and figuratively, is one of our highest priorities. When people form connections, they become a more informed, engaged, and empowered community – individuals with FSHD living a better life, together. An engaged community of patients and families will lead to better understanding of FSHD and faster progress toward treatments and ultimately a cure.

Our staff and volunteer leaders leaned into this theme in their presentations. Gregg Lichtenstein from our Wellness Hour spoke about the value of social capital, and how the benefits of social connection can be intangible, but are immensely powerful (see his essay on page 7.)

Ally Roets from our Early-Onset chapter spoke of the critical need to engage the whole community, not just parents, to ensure our kids are not left behind.

“In a weird sense, I feel grateful to have been diagnosed with FSHD,” Heloise Hoffmann wrote afterward. “It brought me this community that feels like a family: advocating for one another, sharing strengths, leaving no one behind, and perhaps most importantly, fearlessly forging ahead as a rope team despite a daunting mission.”

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# The magic of teams

BY GREGG LICHTENSTEIN, MARGATE CITY, NEW JERSEY

I was once interviewed by a Canadian TV broadcaster, who started by asking how I got into this particular line of work, which at that time involved bringing individual manufacturing firms to work together in groups. And as I began to answer, I could see her eyes go wide, and I could tell she was saying to herself – oh, my God, maybe I shouldn't have asked that question (or – who invited this guy?).



Gregg Lichtenstein

What I told her was that when I was a kid, my favorite comic book heroes were not solo practitioners like Superman, Spiderman, or Wonder Woman; it was the Fantastic Four: a team of specialists with complementary capabilities, who collectively could accomplish what no person could alone.

Why is it that an all-star team (in any sport) is rarely able to beat a championship team? Clearly, an all-star team has better talent at each of the individual positions – better parts or ingredients or building blocks.

The reason is that the relationship or chemistry among the players is the key to the success of the championship team – they know how to *work together*. Chemistry matters, which is why managers and coaches are always looking for the right player to put in their lineup who will catalyze the team to a level of performance that will take them to a championship. In other words, *it is the relationships among the elements* that make a great team more than the sum of its parts.

Did you know that the word



*health* shares the same root as the word *wholeness*? I have come to realize that each of us, no matter what our ability, can only become whole in a community – meaning, in relationship with others. To survive and thrive, we need other people.

What kind of relationships do each of us have with others? In other words, whom do we have on our team? And just as importantly, what kind of relationships do we want and need? Who will carry my water when I need help, and what can I carry for others?

The best communities operate according to the principle “From each according to their abilities to each according to their needs.” As unique individuals we each have something special to offer others, and at the same time we have different needs that others can help us with.

Since its founding in 1991, the

FSHD Society has touched more than 5,000 people around the world who share our particular challenges/abilities, through conferences, programs, and local chapter activities. Increasingly, the Society is focusing on connecting us to each other – in multiple ways (dyads, triads, small groups, large groups, etc.) and on multiple topics (patient challenges, family support, research, etc.) – in order to strengthen our community.

Collectively, we have the opportunity to build teams of all kinds in the FSHD community. We want you to get actively involved. What lights your fire? We only ask you to give that which returns more to you than you put out. What do you need from others, and what do you have to offer them?

We look forward to hearing from each of you. 🙏

# FSHD network grows north

*US\$2 million is invested in Canada to ensure access to clinical trials and treatment*

BY ASIFA LALJI, VANCOUVER, BRITISH COLUMBIA



**A**s a Canadian with FSHD, I am excited – and relieved – to hear about the investment into Canada’s FSHD infrastructure. Like many others in the FSHD community who reside outside the US, I have been following research and clinical trials taking place globally and often wonder if I will have access to a treatment when it becomes available.

While Canadian researchers have been few, they have made significant contributions to FSHD research. However, we have not had the same progress in clinical trials and partnerships with health regulators and pharmaceutical companies as our southern neighbors in the US.

Enter Project Mercury, a global patient-centric initiative launched by the FSHD Society and funded by FSHD Canada Foundation, pharmaceutical companies, and the FSHD Society. In Canada, Project Mercury is focused on expanding clinical trial readiness by adding new Canadian sites capable of conducting pharmaceutical company trials, establishing local FSHD Society chapters nationwide, and accelerating the regulatory process, including reimbursement, for drug and other treatments in Canada.

FSHD Canada Foundation, the only nonprofit in Canada dedicated exclusively to FSHD research, has contributed US\$720,000 to this global initiative. Neil Camarta, co-founder of FSHD Canada Foundation, second-

ed fellow Canadian Ken Kahtava as chief business officer for the FSHD Society to successfully manage this project.

“There is a growing and robust drug pipeline for FSHD right now. For clinical trials to move forward successfully, we forecast the need for at least 10,000 people globally with FSHD who are able and willing to participate in trials when they are available,” said Kahtava. “Without strong patient engagement, and growth of the infrastructure needed to conduct clinical trials with pharmaceutical companies, we can’t move forward.”

The Clinical Trial Research Network, or CTRN, a “center without walls” to conduct FSHD clinical studies, was established in 2016, a few years after a unifying genetic model for FSHD had been established. Researchers Rabi Tawil, MD, and Jeff Statland, MD, realized clinical trial sites and outcome measurements were not in place to take the research to the next level.

The goal was to create research-ready centers along with a toolkit that would provide a comprehensive and open structure for clinical trials for FSHD, including biomarkers, diagnostics, equipment specifications, and data sharing.

The CTRN has grown from four sites in 2016 to 22 sites in seven countries today.

The three new Canadian sites, in Ottawa, Montreal, and Calgary, should be fully functional for both observational studies (like the MOVE study) and interventional trials (like Fulcrum’s REACH study) by mid-2023.



Five additional trial-ready sites are tentatively planned in Edmonton, Vancouver, Winnipeg, Hamilton, and Halifax by the end of 2024. These sites being added in Canada through Project Mercury are not full CTRN sites, but are capable of conducting biopharmaceutical and investigational trials. Setting up these smaller sites in Canada, and globally, is quicker and will help Project Mercury achieve its goals.

A large and engaged patient community in Canada, who are clinical trial ready, will expand the global FSHD patient pool for researchers. This will help pharmaceutical companies move quickly to test promising FSHD treatments.

The progress being made in research and drug development was a driving force behind Project Mercury. Once the drugs have completed the clinical trial process, pharmaceutical companies would need to submit the drugs to regulatory agencies for approval.

Because there is no obligation to submit a drug approved in the US or EU to Canadian health regulators, and an approval in one country doesn't guarantee approval in another country, Camarta realized there had to be a parallel process for regulatory approvals in Canada and other countries.

"We have to be proactive with regulatory bodies in Canada so Canadians will have access to treatment," he said. "This is part of the corporate mission for FSHD Canada Foundation and for me personally."

There is considerable work already in progress to gather data on the FSHD patient community, the financial impact of FSHD on patients and caregivers, and an economic model of the impact of FSHD. This information will help to build the case for clinical trial locations and partnerships, and for biopharmaceutical companies to make the case for drug reimbursement at the provincial level.

The approval process in Canada is a rigorous one, involving federal and provincial regulators, province by province. This takes approximately three years – two for Health Canada to approve safety and efficacy of the drug, and at

least one year at the provincial level to decide on the cost and reimbursement of the drug.

"This project is ultimately all about the patient," said Kahtava. "The nightmare scenario is for there to be a drug or therapy approved in one country and not available in another, or available at an exorbitant cost to the patient. We're doing everything we can to make sure that doesn't happen."


That indeed is a nightmare scenario for more than one million people currently diagnosed with FSHD worldwide.

The central piece for the success of Project Mercury is an engaged patient community. Adding local FSHD chapters will assist in the success of this goal.

The Ontario chapter was first established three years ago. Chapter director Carrie Wilkinson attests to the advantages of local expansion. "Chapters build up the local community and get people ready to participate in clinical trials. They also create a community where people with FSHD can connect and share local resources."

In addition, chapters engage in local fundraising with guidance and support from the FSHD Society. Two Walk and Rolls in Ontario raised nearly \$100,000.

Chapters are also helpful in providing brand recognition and energizing the research community around FSHD. Priscilla Sharun, co-director of the most recently established FSHD Society chapter in British Columbia, is hopeful that merging the existing FSHD community in BC will do just that. "We have a very engaged patient and donor community in BC. We also have world-class researchers, but they're not significantly focused on FSHD. The hope is this activity will motivate them to get involved."

If you are a Canadian with FSHD, it's important to register with the FSHD Society. If you want to participate in clinical trials, please also register with the Canadian Neuromuscular Disease Registry at [cndr.org](http://cndr.org). 

*"...an approval in one country doesn't guarantee approval in another country."*

— ASIFA LALJI



# Your practical guide to clinical trials

BY AMANDA HILL, FSHD SOCIETY

## How do I find clinical trials that are recruiting participants?

Our website is one of the best ways to find clinical trials that are recruiting. We will always have current information for you at [fshdsociety.org/for-patients-families/clinical-trials/](https://fshdsociety.org/for-patients-families/clinical-trials/). Another great way to find clinical trials is at [clinicaltrials.gov](https://clinicaltrials.gov). This website is a database for clinical trials and other clinical research studies all over the world.

To get started on [clinicaltrials.gov](https://clinicaltrials.gov), use the “Find a study” search tool on the homepage. Check the “Recruiting and not yet recruiting studies” status box and enter “FSHD” as the condition or disease. If you want, you can also add your country, state, and city to find clinical trials with study sites near you. Once you get the list of results, you can use filters to narrow your search. Find the “Study Type” filter and check the box for the type of studies in which you are interested.

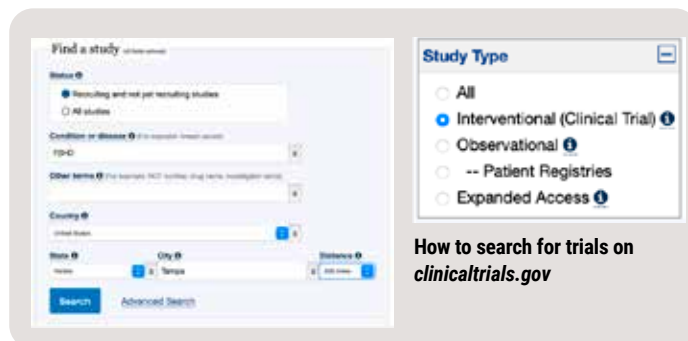
## How do I know if I am eligible for a clinical trial?

You should review the eligibility criteria posted on our website and on [clinicaltrials.gov](https://clinicaltrials.gov) to see if you might qualify for the clinical trial. Some criteria must be assessed by the study team, like your clinical severity score or reachable work-space area. But other criteria you can assess yourself, like your age and health history.

## How do I choose which clinical trial to participate in?

There are many factors to think about when deciding among clinical trials. You should think about the risks and benefits, and about your ability to commit to each study. For example:

- **Are you able to attend all the study visits?** You will most likely have to attend multiple study visits over weeks, months, or even years. Study visits vary in length but can sometimes last an entire day.
- **Will you need to travel?** You may need to travel for some or all of the study visits if there isn't a study site near you. If you need a companion to travel with you, will they be able to take time off from work? Ask if the study can reimburse travel costs for you and/or your travel companion.
- **How much risk are you willing to accept?** The medicines in Phase 1 and Phase 1/2 studies have usually not been tested in people before. These studies can be riskier, and the study team will want to monitor you very closely. This may also mean more work for you, such as keeping a diary of any side effects or going to more study visits.



How to search for trials on [clinicaltrials.gov](https://clinicaltrials.gov)

- **Are you willing to take a placebo?** Phase 2 and 3 studies often compare a medicine to a placebo. This means that some participants do not receive the real medicine. However, you won't know if you are taking the medicine or the placebo until the end of the study.
- **Is there an open-label extension?** Phase 2 and 3 studies sometimes have an open-label extension, which means you may be able to keep taking the medicine for even longer. If you were receiving a placebo during the study, an open-label extension gives you the chance to take the medicine instead.

## How do I sign up to participate in a clinical trial?

1. Use our website or [clinicaltrials.gov](https://clinicaltrials.gov) to find the contact information for the study site you want to go to. This is usually an email address, phone number, or website form.
2. Contact the study site and let them know the name of the clinical trial you are interested in. You might have to wait for the study site to call or email you back. This could take several weeks, but be patient.
3. Usually, the first step in the screening process is a phone call. The study team will tell you more about the study and ask you some basic questions. If they think you will meet the eligibility criteria, they will schedule a time for you to come to the study site for a screening visit.
4. At the screening visit, the study team will talk you through the informed consent. If you agree to participate, the study team will perform all the tests needed to find out if you meet the eligibility criteria. It might take a couple of days to get all the results of your tests back.
5. Finally, the study team will tell you whether you meet all the eligibility criteria. If you meet the criteria, you can choose to continue participating in the study. If you do not meet the criteria, you will not be able to participate.

### What types of activities could I be asked to do during a clinical trial?

You will almost always have to attend study visits in person during a clinical trial, though you may be able to do some visits virtually. The study team may also call you on the phone to see how you are doing or ask you to do certain activities at home. Examples of activities you may be asked to do at study visits or at home include:

- Physical exam.
- Blood draw.
- Electrocardiogram or other tests to monitor your heart.
- Answering questions about your health and ways your disease affects your life. Sometimes, these questions can be very personal. For example, you might be asked about your mental or sexual health.
- Tests to measure your strength, mobility, or range of motion.
- MRI scan.
- Muscle biopsy.
- Taking your medicine at specific times.
- Notifying the study team about any illness, injuries, or side effects you experience.

### Is the medicine safe? What happens if the medicine makes me sick?

One purpose of clinical trials is to learn about the safety of a new medicine. The study team doesn't always know what side effects could happen. This is a risk of participating in clinical trials. However, clinical trials must meet strict standards to help ensure the medicine is as safe as possible.

If the medicine makes you sick, your study team will record what happened. The study team will decide whether they think it is safe for you to continue taking the medicine or if you should stop. They will also help make sure you receive any medical care you need. However, your insurance might have to pay for that medical care. The informed consent form will explain this information.

### Will I learn about the results of the clinical trial?

Yes! You will get to learn about the overall results of the trial, but not your personal scores. However, it could take a long time. Even after you finish your part in the study, other participants might not finish for several more months. The study team must wait for all participants to finish before they can do a full analysis of the data collected. It could take the study

*continued on page 19 ...*

### Glossary:

**Eligibility criteria:** Health traits that allow you to be included in a study, or that cause you to be excluded from a study. These are also called "Inclusion and Exclusion Criteria."



**Informed consent:** The process of learning everything you need to know about a study and deciding if you want to participate. This includes learning about:

- the purpose of the study
- what you will be asked to do
- what tests and procedures will be performed
- the schedule of study visits
- the risks and benefits
- how your data are used

**Intervention:** A medicine or physical treatment that interferes with a disease. Clinical trials are studies that test interventions.

**Open-label extension:** An optional period after the end of the study when all participants receive the medicine, and the study team continues to collect data.

**Placebo:** A mock version of a medicine which looks the same but does not contain the active ingredients that interfere with disease.

**Screening:** A series of questions and tests the study team will perform on you to find out if you meet all the eligibility criteria for a study.

**Study:** A research process to learn about a disease or test a new idea.

**Study site:** A place where a study is performed, often hospitals, clinics, or research centers.

**Study visit:** Defined times when the study team needs to perform tests or collect data from you. Some study visits may be virtual, but many will be in-person.

You will receive a copy of this information in an "Informed Consent Form." If you agree to participate in the study, you will give your consent by signing the Informed Consent Form. Importantly, you can withdraw your consent and decide not to participate in a study at any time and for any reason.

# Ciao Milano 2023!

BY JUNE KINOSHITA, FSHD SOCIETY

This June, the FSHD Society will bring together world leaders in FSHD research and advocacy over five days in Milan, Italy.

Registration is now open to active researchers, clinicians, and research-focused patient advocates.

- June 14: Industry Collaborative Workshop (by invitation)
- June 15-16: 30th Annual International Research Congress (IRC)
- June 17-18: World FSHD Alliance leadership summit (by invitation)




During five intensive days, the FSHD Society will convene stakeholders to chart paths forward in this new era of multiple, global clinical trials. We will welcome attendees to the Radisson Blu Hotel Milan as well as online.

The world's leading thinkers on FSHD will convene for the **30th Annual International Research Congress (IRC)**, expected to draw 400 in-person and virtual attendees from around the world. We anticipate a large number of companies will be there to glean the latest insights on biomarkers and trial outcome measures, as well as to network with key opinion leaders and patient advocates.

The day before the IRC, all of the companies with FSHD programs will be invited to the **Industry Collaborative Workshop**, aimed at evaluating the current status of trial readiness, and identifying opportunities and gaps that could be addressed through collaboration.

On the weekend following the IRC, patient advocacy leaders from 24+ countries will be invited to attend the **World FSHD Alliance leadership summit**. We will share important research advances and updates on global clinical trials and discuss country-specific strategies to advocate for improving healthcare, well-being, and access to treatments for patients everywhere. We will also explore strategies for expanding clinical trial capacity around the world.

Thanks to our benefactors, we are able to offer IRC scholarships and online-only registrations. 



# New frontiers in muscle regeneration for FSHD

BY JAMSHID ARJOMAND, PHD, FSHD SOCIETY

Over the past few years, there has been an increase in the number of pharmaceutical companies developing FSHD therapeutics. These strategies primarily focus on blocking *DUX4*, the toxic element that causes FSHD. These treatments are considered “disease modifying” because they target the root cause of the disease, and if effective, would stop the disease from progressing and becoming more debilitating.

People with FSHD have testified that stopping the disease from progressing is the highest priority. But for the next generation of therapies, strategies are being developed to address the loss of muscle tissue and function. Although this goal was once considered a long shot, there are now several approaches that aim to increase and possibly recover loss of muscle mass, which, if paired with disease-modifying drugs, could in turn reverse the impact of FSHD.

One of these approaches is to boost growth of the existing muscle. This should increase strength and extend functional use of the muscle over the course of the disease. There have already been several trials for this approach in FSHD which have generated mixed results. These include a trial of MYO-029 by Wyeth Pharmaceuticals in 2008<sup>2</sup> and of ACE-083 by Acceleron Pharmaceuticals that concluded in 2020.<sup>3</sup>

While both approaches were deemed safe and showed signs of muscle growth, they failed to show enhanced functional outcomes and were discontinued. However, these trials paved the way for the design of better clinical trials, and in the coming year, Roche Pharmaceuticals will make use of these important findings as they launch a Phase 2 clinical trial for their drug, RO7204239, to promote muscle growth.<sup>4</sup>

In FSHD (and many other muscle conditions), the skeletal muscle gets replaced with fat and scar tissue. This can hinder muscles from rebuilding and contribute to loss of strength. This fat infiltration and scarring is believed to be driven by the immune system. In normal conditions, the immune system helps to maintain healthy muscles. But in FSHD, the constant damage to muscle may overactivate the immune system and release signals that promote scarring and fat infiltration.

Recent FSHD Society grants have invested in basic research to better characterize the type of immune cells and signaling that contribute to fat infiltration and scarring.<sup>5</sup>



This work is still in its early days, but as our understanding of these processes grows, new therapeutic strategies can be developed to block fat infiltration and scarring, and promote healthy muscle regeneration.

Lastly, stem cell therapies are another burgeoning area of research. Most recently, the laboratories of Michael Kyba and Rita Perlingeiro at the University of Minnesota published a study in which mouse stem cells were used to replace the muscle loss in an FSHD mouse model.<sup>6</sup> Although this technique is early in its development, the study highlights the potential for this type of approach and the use of stem cell therapies to rebuild skeletal muscle.

At the forefront of this work is the team from Vita Therapeutics, a Maryland-based biotech company that is developing human stem cell replacement strategies to correct and replace damaged muscles for limb girdle muscular dystrophy and FSHD (see page 14).

All of these innovative regenerative approaches are being developed in concert with disease-modifying therapies to promote a more holistic treatment for FSHD and other neuromuscular indications. The goal is not just to stop disease

*continued on page 19 ...*

# Toward a stem cell-based treatment for FSHD

BY DOUGLAS FALK, VITA THERAPEUTICS

**V**ita Therapeutics is excited to join the effort to develop novel treatments for FSHD, bringing our cellular platform to the fight against this progressive and debilitating disease.

Vita is a biotechnology company located in Baltimore, Maryland, developing state-of-the-art cellular therapeutics for the treatment of neuromuscular diseases. Vita's approach uses induced pluripotent stem cell (iPSC) technology to engineer specific cell types designed to replace those that are defective and causing the underlying problems in patients with genetic diseases. In FSHD, this strategy centers on replacing skeletal muscle cells that secrete the damaging protein DUX4.

VTA-120 is our therapeutic for the treatment of FSHD. It begins with the creation of iPSCs, a process where we re-engineer blood cells from a patient to stem cells, which have the ability to develop into different types of cells, such as muscle. These iPSCs are "treated" in the lab to silence the *DUX4* gene. We then differentiate them into muscle-specific stem cells called satellite


cells. These treated cells, which make up VTA-120, would then be injected back into the patient's muscles, where they would continue to divide and spread throughout the length of the injected muscles, repairing existing damage and ideally growing new, healthy muscle. Because VTA-120 is derived from the patient's own cells, the cells will not be attacked by the patient's immune system, thus allowing repair and regeneration to proceed.

The iPSC technology we are using was originally developed with FSHD Society grant funding by Gabsang Lee, PhD, at Johns Hopkins University, who is scientific co-founder of Vita Therapeutics. Members of the FSHD Society donated blood samples that played a vital role in this pioneering research, which we are now developing into a treatment.

VTA-120 is currently in a preclinical stage, and our aim is to file an Investigational New Drug (IND) application with the FDA for a first-in-human clinical trial in 2025. The development

and success of VTA-120 will build on Vita's first therapy, VTA-100, another autologous iPSC-derived treatment that targets limb-girdle muscular dystrophy type 2A/R1. VTA-100 is currently completing preclinical testing with the goal of filing an IND in 2024.

To help drive our research, Vita is seeking patients with FSHD Type 1 who would be willing to participate in our first-in-human clinical trial. We are seeking individuals in the US with a confirmed genetic diagnosis who would agree to having blood drawn for the purposes of testing the manufacturing process of VTA-120, and who also would agree to non-invasive testing so that we can understand the characteristics of the disease and how the findings would help design an optimal first clinical trial.

If you are interested or would like more information, please email [clinicaltrials@vita-therapeutics.com](mailto:clinicaltrials@vita-therapeutics.com). 



**iPSC-derived muscle cells make healthy dystrophin (green), the protein that is defective in Duchenne muscular dystrophy. A similar strategy might help repair muscles in FSHD. Image: Vita Therapeutics**



# STRENGTHENING OUR WELLNESS MUSCLES

*The value of support groups*

BY ORA PRILLELTENSKY, EDD, AND RANAE BEEKER, MSN RN

“Wellness Hour.” “Women on Wellness.” These are the names of two of the FSHD Society’s mutual support groups, which are the subject of this article. The former, now in its third year and going strong, prompted a request for the latter – an all-women’s offshoot which we fondly refer to as WOW. It’s a cute acronym, but it also underscores a belief that wellness is a common good that can and should be pursued by all, including those who live with challenging conditions and circumstances. Many of the sessions are led or co-led by our own members who have expertise in specific domains, along with lived experience of FSHD.

Living with FSHD presents us with ongoing and progressive challenges to our physical, social, and emotional well-being. Nonetheless, there are actions we are already taking, and some others we can adopt that can improve our quality of life and overall well-being. These are the topics we discuss in our wellness groups. Besides the sharing of knowledge and practical advice about products, procedures, and practitioners, just to mention a few, spending time with others in our community is both meaningful and enjoyable.

Here are some of the topics we covered in our meetings month by month. Managing pain and fatigue; learning about treatment options and clinical trials; sharing our experience with wheelchairs and other assistive devices; managing social and emotional challenges; responding to ableism; doing home modifications; and traveling with a disability. We also spent time connect-

ing in small groups, sharing our stories, and building meaningful connections. This impressive list of topics reflects the wishes and suggestions of our members.

In our beloved WOW group, we explore issues that affect us as women with FSHD. Run and moderated by our very own Ranae Beeker, a retired registered nurse, the group has covered a wealth of topics such as pelvic floor and bladder control, assistance with personal care, body image and self-compassion, dealing with loneliness, feeling like we matter as women with FSHD, and other issues. While the knowledge gained is extremely valuable, the camaraderie and emotional bonds that characterize this unique sisterhood are just as important.

Writing this article is reinforcing our tremendous appreciation for the FSHD Society and for our wonderful members whose expertise, resolve, resilience, and generosity continue to amaze us. We look forward to seeing our regular participants in 2023, and to welcoming new members who can benefit from and enrich our groups with their own gifts.

If you would like to join in on the Wellness Hour or the WOW group, register at [fsbdsociety.org/gathering-place](https://fsbdsociety.org/gathering-place). Additional groups for care partners, parents, young professionals, and “Feeling Fit with FSHD” are also listed on the Gathering Places page of the website. 📌



Ora Prilleltensky



Ranae Beeker

# The Reinventor

A film about coping with constant change

BY AMY BEKIER, SAN DIEGO, CALIFORNIA

How do you describe a progressive muscle-wasting condition and the ever-present elephant in the room, losing independence and the frustrations and fears that follow? How do you relate to patients who feel isolated from family and peers? How can we cope with the constant changes and in some cases, pain, that come with having FSHD?


Jared Jacobsen, the man behind the short documentary *The Reinventor*, does not have FSHD but has family members who do. His production company, Stephen Jake Video, is named in honor of his late father, Dr. Stephen Jacobsen, one of the co-founders of the FSHD Society.

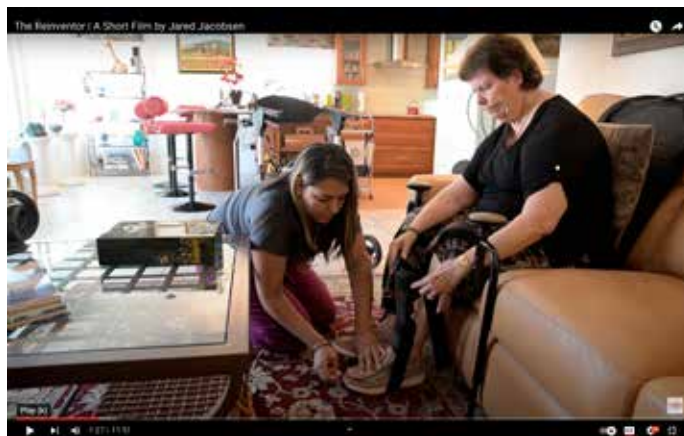
The film's first release was at the Vegas Silver State Film Festival. To date, it has received awards at two of the four festivals where it has been accepted, including best documentary short film at India's Makizhmithran International Film Festival. It is continuing to be submitted worldwide and has been subtitled and dubbed in Spanish.

I met Jared years ago when he volunteered to film local FSHD Society chapter events. He approached me with an idea to create a short documentary about my life with FSHD, and how I cope and maintain a positive attitude. Jared made me feel comfortable during the filming, treating the discussion with creativity and sensitivity. We wanted the same messaging for the film, which was one of clarity, hope, humor, and adaptation.

When I previewed the film, I was surprised at its title, *The Reinventor*. Jared explained that there were numerous times throughout the film when I commented on losing ability and having to "reinvent" myself in search of a worthwhile meaning for my life.

The reaction to this 11-minute documentary both in-person and online has been nothing short of extraordinary. It is being shared worldwide to explain the trials and tribulations of patients and families as they journey through FSHD. It also holds a universal message of optimism, hope, and resiliency that resonates with people who have been touched by other issues as well.

*The Reinventor* can be viewed and shared on the FSHD Society YouTube Channel, [youtu.be/jvd3wKe6S9Q](https://youtu.be/jvd3wKe6S9Q). Jared Jacobsen creator, director, and editor. 



Still of Amy Bekier from the short documentary *The Reinventor*.

# “You’re going to have complex emotions”

BY JARED JACOBSEN, SAN DIEGO, CALIFORNIA



I grew up with FSHD in my household, so I had first-hand experience of living with someone with the disease. It’s a totally different ballgame when you’re young, and later you look back and want to understand it better. In making *The Reinventor*, I wanted to explore this topic further.

My dad was a co-founder of the FSHD Society and a researcher who spent many years studying the disease. I remember him folding up newsletters by hand and mailing them out. I got to know Amy Bekier through local FSHD Society events, and got to see how willing she was to speak about her experiences. I contacted her about being in this film as things were opening up after COVID.

One question that was always interesting to me is the topic of people living with family members who have the disease – the emotions and hardships that come to family members.

That was my personal experience.

Most people have to face the situation at some point of caring for someone who has a serious illness. Amy’s growing up with a parent who had a progressive disease and how she felt about that was an important part of the story.

What stuck out to me is that when she was a kid, she would be angry with her father about the disease. You

I think my dad would be very pleased with how the Society has evolved. That this organization, started more than 30 years ago, is still in operation, and thriving and making such a footprint in the research – he would love that.

Also, he would like the support that the Society is giving to people, patient by patient. He was one of those people who would pick up the phone and always talk to someone. There was no Internet back then, and people would be confused. He was very empathetic, very willing to talk to anyone. He had a lot of knowledge to share, and would be very happy that the Society is filling those roles.

The idea of my film was to reach people, let them know they’re not alone. It has universal themes about moving through hardships. It’s about people having real and serious struggles, and choosing to confront them.

To be a filmmaker, I was told whatever subject you’re attacking,

*“The idea of my film was to reach people, let them know they’re not alone. It has universal themes about moving through hardships. It’s about people having real and serious struggles, and choosing to confront them.”*

— JARED JACOBSEN



have conflicting emotions. You can feel guilt for having these emotions. It’s important for people to realize it’s normal. You’re going to have complex emotions, and you need to forgive yourself for having them.

always think about how I can entertain the audience. I look for someone who is willing to get into the tough subjects, who is knowledgeable, who is a good communicator, who is funny. Amy is all of those things. 🎧

# Adventures with a robotic leg brace

*Stumble control technology reduces falls by 80 percent*

BY KATHY SENECA, NEW ENGLAND CHAPTER LEADER

It can be difficult to find the right adaptive equipment we need to help us navigate the progression of FSHD. My search to stay mobile for as long as possible has led me to the C-Brace.

The Ottobock C-Brace is a long leg brace that uses microprocessor technology to enable the wearer to stand and walk more easily. An integrated sensor system evaluates the stance and swing phase 100 times per second, which the microprocessor uses to control a hydraulic unit.

The brace is individually fitted to each user, and support can be adjusted up or down over the course of time according to your physical needs. The brace responds to your gait dynamically and in real time. Incorporated into this technology is stumble control. Medicare has started covering this brace under strict parameters and documentation, in large part due to a clinical trial that demonstrated dramatic reductions in falls with individuals wearing this device. Also, once the Ottobock Cockpit app is loaded onto your phone, you can access different modes, such as bicycling or adding greater or less support.

Certain criteria must be met to ensure you are a proper candidate for this brace. To be confirmed, there are multiple steps involved prior to receiving the brace. This, in part, includes muscle testing, a trial with C-Brace trial braces, casting, and fittings. After delivery of the brace, physical therapy will assist you with gait training, ramps, steps, uneven ground, etc.

Prior to the C-Brace, I struggled to walk short distances, relying heavily on a rollator. When the brace was delivered, I wore it out of the clinic, and my husband and I immediately enjoyed a quarter-mile walk. It fits like a glove, is wonderfully comfortable, and has given me the freedom and functionality to walk long distances with confidence and ease.

At the time of this writing, I have been wearing the C-Brace for two weeks, and every day look forward to donning the brace and starting my day. Energy level dictates whether I use the rollator or walking poles. We are hopeful insurance will be approving a second C-Brace, and I will become a bilateral user.

As research and clinical trials continue moving forward, exploring the C-Brace may be a helpful option for you to continue managing disease progression. 🙌



Kathy Senecal shows off her C-Brace.



## What's on YouTube

[youtube.com/FSHDSociety](https://youtube.com/FSHDSociety)

### GivingTuesday interviews

Conversations with FSHD rock stars including Kathryn Wagner, Scott Harper, Jeff Statland, Madison Ferris, Max Adler, and many more.

### Feeling Fit with FSHD: Lonwabo Nene's journey

Excellent tips on facial massage, exercises, and more. Videos and downloadable handouts on our blog.

### Feeling Fit with FSHD: Postural Changes

Sessions led by physiotherapist Ulrike Uta of the Muscular Dystrophy Support Centre in Coventry, UK.

### Webinars to watch

All of these webinars are available for your viewing pleasure on the FSHD University page at [fshdsociety.org/fshd-university/](https://fshdsociety.org/fshd-university/).



**Drug Development Update**, with Jeffrey Statland, MD, University of Kansas

**FORTITUDE: A Phase 1/2 Clinical Trial of AOC 1020 in Adults with FSHD**, with Amy Halseth, PhD, and Alissa Peters

**Stem Cell Research**, with Rita Perlingeiro

## Finding your rope team

... from page 6

We each benefit from having others to help us navigate this life. Our volunteer leaders have found their rope team in each other, and they in turn are helping their local communities build theirs. If you haven't found your rope team, reach out to your local chapter. There is no better time. 🙌

## CONFERENCES



### 360 Conferences

In person. Videos available post-conference.

**May 6:** University of Rochester, New York

**October TBD:** University of Washington, Seattle

**November 11:** The Ohio State University, Columbus

## WEBINARS



### FSHD University webinars

Advance registration required.

**March 16:** Novel therapeutic approaches, Nizar Saad, PhD

**April 20:** Phase 2 clinical trial in FSHD, Roche Pharmaceuticals

**May 18:** What every newly diagnosed person should know, Bakri Els Sheikh, MD

**July 20:** Insights from Europe: FSHD patients' thoughts on clinical trials, Megan McNiff, PhD

## FSHD SOCIETY RADIO



Community Profiles, second Tuesday of each month, are in-depth interviews with remarkable people in our community. Hot off the Press, fourth Thursday of each month, is short-form shows that will bring you the latest and greatest from the FSHD Society. All episodes premiere at 9 p.m. ET | 8 p.m. CT |

7 p.m. MT | 6 p.m. PT on the dates listed, and are available to stream on-demand on major streaming platforms.

## CHAPTER MEETINGS



Our 30+ chapters are busy planning meetings for this year, 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



### Early-Onset Parent Roundtable

Third Tuesday of every month at 8 p.m. ET | 7 p.m. CT | 6 p.m. MT | 5 p.m. PT. One-time registration required.

**March 21**

**April 18**

**May 16**

**June 20**

**July 18**

### CarePartner Hour

Last Tuesday of every month at 8 p.m. ET | 7 p.m. CT | 6 p.m. MT | 5 p.m. PT. One-time registration required.

**March 28**

**April 25**

**May 30**

**June 27**

**July 25**

### Wellness Hour

Second Monday of every month at 5 p.m. ET | 4 p.m. CT | 3 p.m. MT | 2 p.m. PT. One-time registration required.

**March 13**

**April 10**

**May 8**

**June 12**

**July 10**

### 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. One-time registration required.

**April 5**

**May 3**

**June 7**

**July 5**

### Feeling Fit with FSHD

Our newest Gathering Place meets on the second and fourth Thursday of every month at 6 p.m. CET | 5 p.m. GMT | noon ET | 11 a.m. CT | 10 a.m. MT | 9 a.m. PT

**March 9 & 23**

**April 13 & 27**

**May 11 & 25**

**June 8 & 22**

**July 13 & 27**


**Visit our Events Calendar for updates and to register for events ([fshdsociety.org/fshd-events-calendar](https://fshdsociety.org/fshd-events-calendar)).**



## Your practical guide to clinical trials


... from page 11

team 6-18 months to perform the analysis and prepare to share the results. Sometimes, a study team will share preliminary results. Often, the FSHD Society will schedule a webinar to explain the results to the community.

Many study teams will notify participants when they have results to share, for example, through an email newsletter. Sometimes, you will have to watch out for news stories, social media posts, or a press release. Most study teams will also publish their results in scientific journals or present them at scientific conferences. 

## New frontiers of muscle regeneration for FSHD

... from page 13

progression, but to find treatments that can someday reverse the damage and restore functions that have been lost. 

<sup>1</sup> Voice of the Patient Report: [fshdsociety.org/fsh-events/vopf/](https://fshdsociety.org/fsh-events/vopf/)

<sup>2</sup> Please see page 4 of FSHWatch at [fshdsociety.org/wp-content/uploads/2019/08/FSHSociety\\_FSHWatch\\_Spring2008.pdf](https://fshdsociety.org/wp-content/uploads/2019/08/FSHSociety_FSHWatch_Spring2008.pdf)

<sup>3</sup> [fshdsociety.org/2019/09/16/disappointing-news-from-acceleron/](https://fshdsociety.org/2019/09/16/disappointing-news-from-acceleron/)

<sup>4</sup> [clinicaltrials.gov/ct2/show/NCT05548556](https://clinicaltrials.gov/ct2/show/NCT05548556)

<sup>5</sup> Studies by Yegor Vassetzky & Carlo Serra at [fshdsociety.org/grants/grants-we-have-funded/](https://fshdsociety.org/grants/grants-we-have-funded/)

<sup>6</sup> [pubmed.ncbi.nlm.nih.gov/36056021/](https://pubmed.ncbi.nlm.nih.gov/36056021/)

## New opportunities, new challenges

*The most exciting – and urgent  
– era yet in FSHD history*



This year, we have entered a new phase in drug development

- For the first time ever, there is a drug in Phase 3 of a clinical trial – the final phase before seeking FDA approval.
- For the first time ever, there will be multiple formulas in clinical trials simultaneously, meaning that you could have multiple options for volunteering.
- These milestones, along with so many others, give us greater hope than ever of delivering treatments to families in the near future.

You, and generous donors just like you, made this happen. Now, as we embrace new opportunities, we also face new challenges. Now, we are being called on to become more.

By donating to the Spring Campaign, you will help speed up the development of treatments. Your gift will amplify the voice of the FSHD community, ensuring the regulators hear us loud and clear. Even better, your impact will be doubled thanks to matching gifts pledged by our Matching Gift Circle of donors. *In this new era, your support is more important than ever.*

### WAYS TO DONATE:

- Scan the QR code
- Go to [FSHDSociety.org](https://FSHDSociety.org)
- Or mail a check to:  
FSHD Society, Department 960,  
PO Box 4106, Woburn, MA 01888 USA

