

FSH Watch



CONNECTING THE COMMUNITY OF PATIENTS, FAMILIES, CLINICIANS, AND INVESTIGATORS



FSH Society's 2016 International Research Conference

Workshop highlights growing focus on translational research for FSHD

by CHARIS HIMEDA, PhD
Reno, Nevada

Leaders in FSHD research joined clinicians and industry partners at the FSH Society's 2016 International Research Consortium and Research Planning meetings held in Boston on November 10-11, 2016. Building on momentum gained over recent years, the workshop emphasized translational studies geared toward identifying treatment targets for FSHD and the steps necessary to bring future therapies to the clinic.

There is now a general consensus in the field that FSHD pathology is caused by a loss of DNA repression at the disease

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Researchers from around the world attended the 2016 FSHD Research Connect conference in record numbers.



Two Lovelies greet guests arriving for the 2016 Ghostly Gala at the legendary Cicada Club in downtown Los Angeles. The FSH Society's annual Halloween costume gala was live-streamed.



Young Hollywood steps up for A Ghostly Gala

People Magazine and Perez Hilton tweet to 13 million followers!

by ALISON KORMAN
Pasadena, California



The FSH Society celebrated its second annual Ghostly Gala to Vanish FSHD at the historic Cicada Club in downtown Los Angeles on Sunday, October 30, 2016. With the theme "Bach to Bowie: Music Through the Ages," the event honored Mimi Garcia

and her family for their years of dedicated service and generosity to the FSH Society and community. It was a magical night filled with music, dinner, dancing, silent and live auctions, and a costume contest judged by a hot new crop of young Hollywood actors.

The Host Committee—Co-Chairs Trish Doktor and Alison Korman, and members Ellen Rennell, Selina Lai, Chareen Kossoff, Laurie Heyman, Stephen O'Connor, David Garden, and Rachael

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Against all odds, dreams fulfilled

Dear Friends,

Our president and CEO, Dan Perez, began his work of founding the Society in the context of Stephen Jacobsen's efforts to identify large families with FSHD and establish cell lines for genetic studies to pinpoint the cause of FSHD. Stephen was co-founder of the FSH Society and a patient himself who worked as a researcher on FSHD. He passed away in January of 2006, a tremendous loss not only for his family and Dan, but for the entire FSHD community.

Many of the Society's founding families contributed blood samples for this work. Over time, 607 samples were collected across 43 families. Through a series of events, this research ran out of funding. One copy of the collection was moved, one of the libraries completely lost, and the surviving library was partially rescued but was locked down in deep freeze at a national repository due to inadequate information to allow it to be made available for research.

It gives me incredible joy to report that this invaluable resource has been liberated, thanks to herculean efforts by Dan and the laboratory of Peter Jones, and is available to researchers through the Coriell Institute for Medical Research, a "biobank" supported by the National Institutes of Health. You can learn the details in our story on page 3.

Through this achievement, we honor the memory and life's work of Stephen Jacobsen. The FSH Society is deeply grateful to the Jones lab and the biobanking leadership at the Coriell Institute, for their whole-



June Kinoshita

hearted dedication to completing this project, and to the individuals and families who donated specimens all those years ago.

Many other dreams are also being fulfilled. A Dutch study reported that exercise and cognitive behavioral therapy may slow down and even reverse muscle degeneration in FSHD patients (page 4). What patient hasn't dreamed of this? This is just a single study so far, but such research gives us great hope.

George Pollock is pursuing his life dream to ride in the Race Across America, and his eight-person team will dedicate its effort to raising awareness and funds for the Society (page 15). For Trisha Sprayberry, the dream was to travel independently with her wheelchair to attend her first FSHD patient conference (page 10).

For myself, I dreamed of bringing individuals from across the FSHD community—patients, families, researchers, and donors—together at FSHD Connect in Boston last November to honor Dan Perez and other heroes (page 16), celebrate our strength and accomplishments, and rededicate ourselves to fulfilling our dreams for all those living with FSHD.

The hard work and generosity of our Board of Directors, Scientific Advisory Board, staff, volunteers, and donors make all of this possible. We are profoundly grateful for your support.

Sincerely,



June Kinoshita
Executive Director & Chief Operating Officer

FSH Watch

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It is our editorial policy to report on developments regarding FSHD, but we do not endorse any of the drugs, procedures, treatments, or products discussed. We urge you to consult with your own physician about any medical interventions.

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New source for FSH muscular dystrophy family cell lines

Thirty-year-old effort by FSH Society finally sees light of day

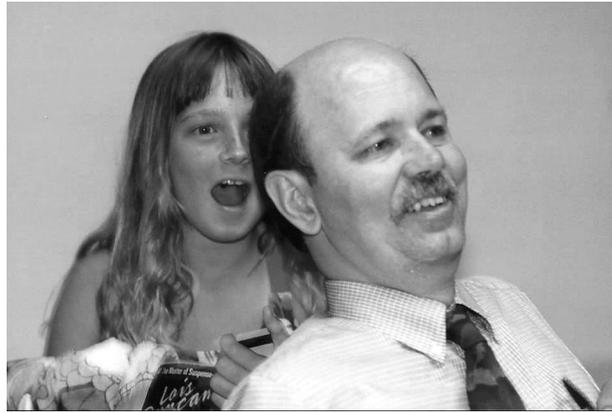
by **JUNE KINOSHITA**
FSH Society

FSHD researchers now have an invaluable new resource for understanding and developing treatments for FSHD. An important collection of cell lines, comprising cell lines from 114 individuals with FSHD representing 12 multigenerational families, are being made available through the National Institutes of Health Human Genetic Cell Repository at Coriell Institute for Medical Research.

This work, begun in 1987 by Daniel P. Perez and the late Stephen J. Jacobsen, PhD, fellow FSHD patients and co-founders of the FSH Society, represents the culmination of a 30-year journey to generate much-needed resources for the FSHD research community. These cell lines were derived from clinical samples taken from FSHD patients and their unaffected relatives between 1987 and 1992, based on the clinical assessments of Dr. Jacobsen and other neurologists with expertise in the disease. There was no genetic test available for the disease back then.

Unfortunately, information required to allow public distribution of these cell lines was not available at the time. The cells were warehoused in a deep-freezer at the Coriell Institute, preserved but not able to be released to the research community. Over the past two and a half years, Peter and Takako Jones' lab at the University of Massachusetts (now at the University of Nevada, Reno School of Medicine) collaborated with the FSH Society and the Coriell Institute to confirm the pedigrees by short tandem repeat (STR) analysis and produce critical information relevant to FSHD.

The cell lines were characterized to provide the data describing the multiple genetic features associated with the disease. Specifically, each cell line was characterized at the FSHD region of chromosome 4 for the D4Z4 array proximal simple sequence length polymorphism (SSLP) and array distal subtelomere haplo-



The late Stephen J. Jacobsen, PhD, with his daughter.

type, tested for FSHD1 and FSHD2 genetics and epigenetics, and assayed with respect to expression of DUX4, a key gene implicated in the disease. In addition, a subset of cell lines was confirmed to express DUX4 target genes and shown to be responsive to epigenetic drug treatments, indicating their suitability for use in testing therapeutic approaches targeting DUX4 expression.

With this initial characterization, these cell lines are now available through the Coriell online catalog (<https://catalog.coriell.org/1/NIGMS/Collections/Heritable-Diseases>). The

NIH Human Genetic Cell Repository at Coriell is sponsored by the National Institute of General Medical Sciences (NIGMS).

Perez, who serves as president, CEO, and CSO of the FSH Society and is a senior corresponding author on the study, commented, "Through this work we honor the memory and life's work of an old and dear friend. The FSH Society is indebted to the Jones lab and the biobanking leadership at the Coriell Institute for accomplishing this formidable task, and to the individuals and families who donated specimens all those years ago. It is a privilege to help make these biomaterials openly available to all communities pursuing research and clinical efforts on FSHD. I look forward to seeing what new insights and progress will come from our efforts."

Investigators looking to purchase the cell lines should have a current MTA on file with the NIGMS Repository at the Coriell Institute. Complete information on how to order samples can be found at this link: <https://catalog.coriell.org/1/NIGMS/How-to-Order>. 

Reference

Takako I. Jones, Charis L. Himeda, Daniel P. Perez, and Peter L. Jones. 2016. Large family cohorts of lymphoblastoid cells provide a new cellular model for investigating facioscapulohumeral muscular dystrophy. *Neuromuscular Disorders*. 23:975-980.

Family members of FSHD patients needed for study

IF YOU HAVE NO SYMPTOMS, YOU MAY QUALIFY

Want to help further FSHD research for your affected family members? The Johns Hopkins Hospital and the Kennedy Krieger Institute are seeking participants for a new study of FSHD relatives. They are seeking near relatives of FSHD patients (parents, children, or siblings) over the age of 35 who are not currently showing symptoms.

There is no need to be living in the Baltimore area

to participate in this study. The participation in the study consists of a blood draw which can be performed at any local lab near your home. All accompanying costs, including a genetic test, will be covered by the study.

For more information, please contact Pegah Dehghan (dehghan@kennedykrieger.org). The study's protocol number is NA-00019985.



Therapies appear to slow muscle degeneration

Imaging study shows positive effects of exercise and cognitive behavioral therapy

by **KATHRYN PUZZANGHERA**
FSH Society

In 2014, a Dutch team reported that aerobic exercise training (AET) and cognitive behavioral therapy (CBT) decreased fatigue and improved the quality of life significantly in FSHD patients. Now, the same group has published a study demonstrating that not only did patients given AET or CBT feel more energized and active, but that their muscles degenerated more slowly than in patients who received standard care.

Strikingly, the effect was largest in the CBT group. CBT often focuses on how your thoughts can influence your behaviors and the choices you make. It is often used to treat patients with chronic illness to improve their functioning in their daily life.

The study

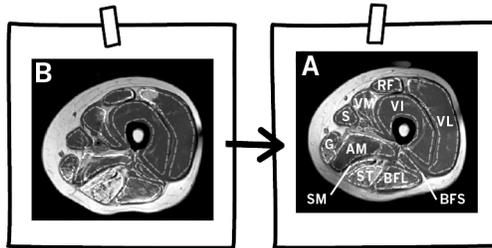
The study comprised 31 patients in total; 13 received standard care, nine received AET, and nine received CBT. The results were primarily measured via magnetic resonance imaging (MRI). The scientists looked at edema and fat infiltration in the muscles. Edema is the level of water in muscles, and is interpreted as a sign of inflammation. When muscles degenerate, they are replaced by fat. This process is known as fat infiltration, and the fraction of a muscle group that has turned to fat is a good indicator for the progression of the disease.

With the MRIs, the scientists were able to measure the percentage of fat in a muscle and draw conclusions about the progression of the disease. They noted that this is a very useful metric that should be utilized in future studies.

The results

While not every muscle responded the same to the treatments, the study showed significant differences between the control group of untreated patients and the patients who received either CBT or participated in an aerobic exercise program. In the control group, fatty infiltration progressed 6.7 percent over the course of a year. However, in the aerobic group, the increase was only 2.9 percent, and in the CBT group only 1.7 percent. This suggested a marked impact in slowing the degeneration of affected muscles.

Beyond these measurable physical effects, the patients who received CBT or AET showed significant improvement in their physical activity, which led to, among other things, a decrease in their level of fatigue, giving them the ability to participate more fully in their activities and improving their quality of life.



Magnetic resonance image (MRI) shows the cross-section of the upper leg of an FSHD patient. Image A (right) was taken 28 weeks after image B (left) in a patient who received cognitive behavioral therapy. The fat infiltration (white) decreased, especially in the quadriceps (vastus lateralis, VL) and hamstring muscles (semimembranosus, SM).

“ It is important to become, or remain, physically active because physical activity has a positive effect on fatigue, well-being, overall health, and fatty infiltration. ”

NICOLINE VOET, MD

What might be going on?

According to study co-author Nicoline Voet, MD, “We think that an increase in physical activity is essential to treat fatigue and decelerate the increase of fatty infiltration in muscles of patients with FSHD.”

She noted that an essential goal for CBT is to increase physical activity. CBT also addresses ways to manage fatigue, pain, sleep disorders, and psychological barriers to participating in social activities. The resulting increase in physical activity, Voet hypothesizes, might lead to “epigenetic changes and a decrease in inflammation,” which could be the underlying mechanism that slows down the rate of fatty infiltration.

Future directions

The Dutch group would like to see researchers in other countries replicate their study, Voet said.

This year, the Dutch group plans to recruit patients for a new trial, called “Life Balance,” which aims to help patients

balance their current and future activities with their mental and physical capacities. Part of the plan for this trial is to replicate the CBT study and investigate underlying epigenetic factors, said Voet.

What does this mean for you?

“It is important to become, or remain, physically active,” Voet suggested, “because physical activity has a positive effect on fatigue, well-being, overall health, and fatty infiltration.”

The study suggests that CBT can be effective in encouraging greater physical and social activity in FSHD patients. It also suggests that FSHD patients are likely to benefit by incorporating aerobic exercise into their daily routines.

It is important that you consult with your doctor or physical therapist to make sure you find the right balance for your body, and it is advisable to try a graded exercise plan (starting slowly, maybe with only a few minutes, and working yourself up in small increments every day). [FSH Watch](#)

Reference

Barbara H Janssen*, Nicoline B M Voet*, Alexander CH Geurts, Baziel GM van Engelen, Arend Heerschap. Quantitative MRI reveals decelerated fatty infiltration in muscles of active FSHD patients. *Neurology*. 2016;86(18):1700-7.

*contributed equally.

FSHD imaging study seeks volunteers

Early-stage patients of particular interest

by JUNE KINOSHITA
FSH Society

The Kennedy Krieger Institute is recruiting volunteers with FSH muscular dystrophy for a clinical research study. Volunteers will be asked to undergo muscle strength testing and non-invasive magnetic resonance imaging (MRI) and spectroscopy (MRS) of the muscles. The strength testing and MRI/MRS scans will be repeated every three to six months for up to two years.



Doris Leung, MD

The study is particularly interested in recruiting individuals over the age of 12 years old who started having symptoms of FSHD within the past three years.

Although there are no direct benefits to volunteers who participate in this study, the information collected will be used to develop disease biomarkers for FSHD.

“Many of our patients with FSHD tell us that they started developing symptoms in adolescence or (looking back) feel that there was a period of more rapid progression around this time,” said Doris Leung, MD, the principal investigator of the study. “It’s unclear to us why this happens, and we’d like to learn more by following a group of these patients closely over time.

“We’re going to be collecting detailed medical histories and performing strength testing along with the sequential MRI scans, and this could give us greater insight into factors that trigger disease progression,” Leung explained.

Leung thinks the research could help individuals reach a correct diagnosis sooner. “I suspect that the diagnosis of FSHD is frequently missed at the beginning,” she said. “By the time patients see us [at the Kennedy Krieger Institute] for the first time, many have already gone through a long diagnostic odyssey that involved seeing multiple physicians and undergoing potentially unnecessary tests.

“Although it’s not a direct aim of the research, I’d like to think that having a better characterization of what FSHD looks like in the early stages can indirectly raise awareness among physicians and lead to patients being diagnosed faster,” Leung said.

Individuals interested in volunteering, should

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Ask Beth

FINDING FREE MONEY FOR THE CAUSE LOOK NO FURTHER THAN YOUR EMPLOYER!

Raising awareness and funds for a rare disease that few have heard of can be an uphill battle. With this issue of the *FSH Watch*, we are launching a regular advice column by our own Beth Johnston, who answers frequently asked questions about addressing these challenges. Do you have a question or suggestions? Write to Beth at beth.johnston@fshsociety.org.



Beth Johnston

Q. I support the FSH Society by donating what I can afford, but I wish I could do more. I've heard that some companies match charitable gifts made by their employees. How do I take advantage of this “free money”?

A. Yes, there's free money out there. It's called company matching gift programs, which are an excellent way to maximize the value of your donation!

Many companies have matching gift programs that are designed to support and encourage employee charitable giving. Companies big, small, and everything in between, match gifts.

Approximately two-thirds of companies with matching gift programs match employee donations to nearly any 501(c)(3) organization or educational institution.

The other one-third of companies match donations to specific types of organizations, such as:

- educational institutions (K-12 included);
- health and human services;
- arts and cultural organizations;
- civic and community organizations; and
- environmental organizations.

Other companies support employee volunteer activities. Take the case of Johns-Manville in Littleton, Colorado. Jeanne Virtue, a committee member on our 2016 Walk & Roll to Cure FSHD in Castle Rock, Colorado, and an employee of Johns-Manville, organized a group of 20 employees to participate in the walk. Johns-Manville covered their entrance fees, which became a donation of \$1,000! Everyone showed up in matching shirts, had a great time, and raised money for the FSH Society. Thank you, Jeanne, and Johns-Manville!

What can you do? Check with your human resources office to see if your company has a matching gift program. If it does, request the form. Review the guidelines to see if the FSH Society is eligible. Mail the completed form to the FSH Society, ATTN:

Robyn O'Leary, 450
Bedford Street, Lexington,
MA 02420. We'll take care
of the rest! Your company
will send the matching
gift directly to the FSH
Society.



Employees from Johns-Manville turned out in force to support the 2016 Castle Walk & Roll to Cure FSHD.

Intriguing research on tyrosine kinase inhibition as a potential therapy for FSHD family cell lines

Sunitinib rescues muscle cells' ability to develop

by **JIM ALBERT**
Eldersburg, Maryland

A cancer drug has been shown to potentially rescue some of the damaging effects of DUX4, the gene implicated in FSH muscular dystrophy. The laboratory of Peter Zammit, PhD, Randall Division of Cell and Molecular Biophysics, King's College London, United Kingdom, in collaboration with Robert Knight, PhD, of the Department of Craniofacial Development and Stem Cell Biology at King's, has published the results of its research on the activity of an FDA-approved drug, sunitinib, as having potential therapeutic activity for FSH muscular dystrophy (FSHD).

This research, sponsored by Muscular Dystrophy UK, the FSH Society, and the French Muscular Dystrophy Association (AFM-Téléthon), arose from the finding that the gene RET (Rearranged During Transfection) was upregulated by the DUX4 protein in mouse muscle cells, as well as from work investigating the importance of RET in controlling the stem state of muscle progenitor cells in zebrafish.

While this research is at a very early stage and requires much further work, it is encouraging as it shows the potential for existing drugs to point to new strategies in the treatment of FSHD.

Sunitinib is approved for the treatment of certain types of cancer and inhibits cellular signaling by targeting several cell receptors, one of which is RET, a receptor tyrosine kinase. RET is activated by small proteins released by other cells, and until now has not been shown to be involved in adult muscle biology. This collaborative study revealed that RET is expressed in muscle stem cells and is required for their proliferation, indicating RET acts in the normal control of skeletal muscle formation and repair.

The overexpression of the DUX4 gene, or more properly DUX4-fl (full length), is widely believed to be the catalyst that sets off a chain of events leading to FSHD. While the research of the Zammit and Knight team showed that RET normally

plays a beneficial role in muscle, in the presence of DUX4, RET production was increased, which can adversely affect muscle cell differentiation, the process of muscle stem cells forming skeletal muscle fibers. In tests performed in this research, sunitinib suppressed RET signaling to rescue DUX4-mediated inhibition of muscle differentiation in DUX4-expressing stem cells.

One observation made in this work revealed that RET is beneficial to healthy muscle formation, unless deregulated, when it is potentially detrimental. What might that mean in terms of attacking RET signaling as a therapeutic avenue in FSHD? Zammit answered this by commenting, "RET has a role in normal muscle formation in adults. Further research might focus on reducing levels from the abnormally high levels of RET signaling caused by DUX4 to more normal levels."

The results of the Zammit and Knight team's research in mouse cells were validated both in vitro and in vivo using muscle cells from an FSHD patient. The in vivo studies were performed by grafting the patient's FSHD myoblasts into regenerating muscles of immunodeficient mice (lacking a normal immune system, so as to prevent immune rejection of the foreign cells).

Suppressing RET activity using sunitinib rescued the ability of both DUX4-expressing mouse myoblasts and FSHD patient-derived myoblasts to differentiate, or develop, into muscle. Sunitinib application also aided the engraftment and muscle cell differentiation of FSHD patient myoblasts that had been transplanted into mice.

This research showed that DUX4-mediated activation of RET contributed to the inhibition of myoblast differentiation and so might contribute to FSHD pathology by preventing stem cell-mediated repair of muscles. What's more, the study showed that sunitinib rescued DUX4-induced pathology involving muscle cell differentiation, suggesting the therapeutic potential of tyrosine kinase inhibitors for the treatment

of FSHD. (See figure on page 20.)

The researchers reported, however, that sunitinib does not prevent muscle cell apoptosis ("programmed cell death"), which may also play a role in damaging muscles in FSHD.

It is important to note that while research on RET signaling and FSHD is interesting, an increase in RET signaling is only one of the many consequences of DUX4 expression in muscle progenitor cells. Zammit commented, "RET is one of a plethora of changes that DUX4 causes in muscle. Countering RET effects leaves all these other changes in place, and they are still likely to be detrimental. Stopping DUX4 is probably the best option, as it stops all the downstream effects, too."

While stopping DUX4 is indeed the leading contender for FSHD therapy, many doctors and scientists believe it may eventually be a cocktail of therapies which proves to be the best overall treatment of FSHD. This is especially true if one of the initial DUX4-targeted therapies only slows the expression of DUX4 rather than turning off DUX4 completely.

While it is encouraging that an already approved drug could possibly be part of such a treatment for FSHD, sunitinib does come with a variety of potentially serious side effects such that, while it may be a reasonable treatment for a severe form of cancer, the risks might not be appropriate for FSHD.

Zammit further commented, "While the data from this research is preliminary, uncovering the targets of sunitinib may identify targets that can be countered with safer drugs, or there might be a version of sunitinib appropriate for FSHD that has fewer side effects. Sunitinib, or another tyrosine kinase inhibitor, may not be a solo therapy but could be useful in combination with other drugs that target other aspects of the pathology, such as apoptosis."

In response to questions about plans to expand on this research, Knight replied, "Our next step is to understand how sunitinib

tinib affects gene expression in FSHD patient-derived myoblasts. We have already started on this with the aim to identify candidate target molecules that we can focus on in more detail. In parallel, we intend to perform a screen for proteins showing altered activity in these cells in response to sunitinib, focusing on receptor tyrosine kinases.” [FSH Watch](#)

Reference

Louise A Moyle, Eric Blanc, Oihane Jaka, Johanna Pruessler, Christopher RS Banerji, Francesco Saverio, Tedesco Stephen, DR Harridge, Robert D Knight, Peter S Zammit. Ret function in muscle stem cells points to tyrosine kinase inhibitor therapy for facioscapulohumeral muscular dystrophy. *eLife*. 2016;5:e11405. Online at: <https://elifesciences.org/content/5/e11405>.

CHARITY NAVIGATOR AWARDS THE FSH SOCIETY WITH HIGHEST RATING

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More than 2,000 donors from around the globe contributed to the Society in 2015 (the year on which the current Charity Navigator evaluation is based), with more than 95 percent of total revenue coming from individual patients, family members, and their friends. Most importantly, 84 percent of the Society's budget was invested in programs to promote the health and well-being of patients and families through education and research toward more effective treatments and a cure.

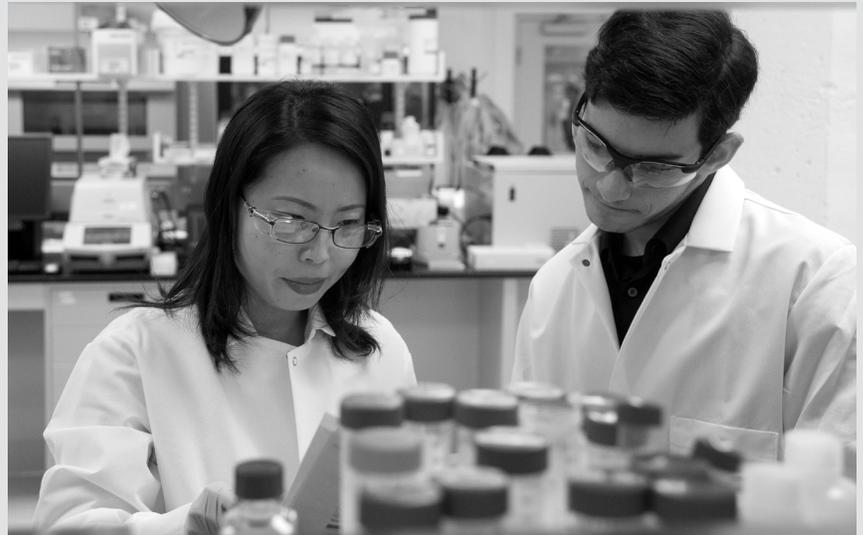
We are so grateful to our donors for their loyalty and generous support! [FSH Watch](#)

FSHD IMAGING STUDY SEEKS VOLUNTEERS

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contact Doris Leung, MD, at (443) 923-9521 or email her at leungd@kennedykrieger.org.

This study is funded by an NIH K23 Mentored Patient-Oriented Research Career Development Award and is conducted under Kennedy Krieger Institute, Johns Hopkins Medical Institutions Study Protocol Number NA_00065256. [FSH Watch](#)



Researchers at Fulcrum Therapeutics will use cell lines derived from tissue donated by an FSHD patient who had surgery.

DONATED PATIENT TISSUE IS HELPING TO ADVANCE RESEARCH

At Fulcrum Therapeutics, human tissue serves as one of the most basic yet essential tools available to help in efforts to develop new medicines to treat FSHD and other genetic diseases.

Human biospecimens have long served as a foundation for the development of precision medicines. By deeply analyzing human tissue at the cellular level, researchers gain indispensable insights into how a disease progresses, which may open the door to new treatment strategies. These insights also enable the development of personalized molecular tools that are used to evaluate the safety and efficacy of novel therapeutics as they move through human clinical trials.

The FSH Society has partnered with the National Disease Research Interchange (NDRI) to establish a US-wide registry of individuals with FSHD and their family members who wish to donate tissue obtained during surgery or postmortem. The registry obtains informed consent from donors, and arranges to recover donated tissue and distribute it to qualified researchers, including Fulcrum Therapeutics.

Using this muscle tissue, Fulcrum has developed FSHD primary muscle stem cells known as myoblasts which are capable of being expanded and cultured in our laboratories for many uses. Fulcrum will use these FSHD myoblasts for disease characterization and early drug discovery activities. In addition, Fulcrum researchers are using these important tools to ask further questions, including: What unique markers of disease can be found in these tissues? How do these markers affect normal muscle function? Can new tests that measure these unique markers of FSHD be developed? Can novel therapeutics that impact disease characteristics of FSHD be identified?

The FSHD Biospecimen Registry has proven to be an invaluable resource, and the tissue samples obtained are helping enormously in Fulcrum's efforts to deliver a new future for patients and their families. Fulcrum Therapeutics is sincerely grateful to patients willing to participate in tissue donation, and to the FSH Society for their collaboration.

— Kelly Jackson
Fulcrum Therapeutics
Cambridge, Massachusetts

Accessibility hacks, mobility aids, and tips

Useful tricks for coping with weakening muscles

by PAUL SHAY
Groton, Massachusetts

I am one of the lucky, late-onset FSHers: diagnosed at age 57, now 71. As my progression has been relatively slow over those 14 years, I have incorporated a number of adaptive devices and techniques that I can share with you. Since my wife and I are still working, we have been fortunate enough to have the resources to try out a number of products.

As I am mostly affected in my shoulders, biceps, and quads, my issues revolve primarily around climbing stairs, reaching high and low, and getting up from a seated position.

For those challenges, here are a number of things and tricks I've found useful:

1 PNEUMATIC CLAM-SHELL SEAT LIFT

This device is adjustable for its user's weight and has modest padding. It allows me to sit at my desk for hours at a time, and gives a gentle boost when getting out of the chair.

2 TOILEVATOR

This is a sturdy plastic platform onto which your toilet mounts. It comes with all necessary mounting hardware and seals, raising toilet seating height an additional four inches.

3 HALF-STEPS

We have a garage with one step up to the porch, and inside, a single step from the entry area to the main level of the house. When these became an issue, I tried plastic half-steps purchased online. They proved the concept and did help, but were fairly expensive and yet did not last long. Our son then built a couple of rectangular boxed platforms of the same height using quality hardwood. After staining them, he added 3M non-skid strips for traction.

For travel, we bought a 12-inch square stepstool and cut the legs off to bring the height down to that of our household half-steps. It fits easily in a suitcase or carry-on, and takes up minimal space.

Otherwise, one of my early purchases was a cane with a small half-step attached to the base. In conjunction with my normal cane, most steps and stairs are now doable thanks to this device.

4 GRAB BARS

In conjunction with my half-steps, we added a conventional grab bar in the doorway to our garage. My wife came up with the idea of using a six-inch, black wrought iron drawer pull in the doorway in our house that contains the step from the entry area to the main level. It works very well, and it looks much less obtrusive and "institutional" than a standard grab bar.

For our shower, we purchased a couple of 12-inch suction cup-



This folding toilet frame has been one of our best purchases. It is fully height adjustable and fits behind the seat of any commode without mounting hardware. We can take it on trips.

mount grab bars and installed them on the walls. They are intended only for assisting in stepping into and out of the shower, and for help with balance—they can't bear your full body weight. The best thing about them is their portability, thanks to the suction cup-mounting system. They fit handily into a suitcase and make it possible to use hotel rooms that lack installed mobility aids in conjunction with:

5 FOLDING TOILET FRAME

Essentially a portable set of armchair-like padded rails, this has proven to be one of our best purchases. It is fully height adjustable and fits behind the seat of any commode without mounting hardware. We have been able to toss it in the trunk and take off on trips without having to make an ADA-compliant room reservation.

6 STAIRLIFT

By far our largest purchase to date has been the installation of a stairlift in our home. With our master bedroom and my office on the second floor, access was becoming a serious issue. We were

able to buy a gently used unit with a factory warranty directly from Stannah for \$4,300 (including the use of an Angie's List coupon). While that is not an insubstantial amount, it beat having to move! The unit has a battery backup feature in case of a power failure, and remote controls that allow the chair to tote bulky/heavy objects up and down the stairs.

7 REACHER-GRABBERS

I have four of these—one for the garage (I've dropped my keys more than once), two for the main living area of our house, and one for upstairs. Three of them are "cheap-o" models I got for about \$4 each at Harbor Freight Tools. The fourth is more substantial, with rotating grip jaws and a telescoping body. It is robust enough that I can lay a fire with it, including placement of small logs.

MORE FUNDING FOR RESEARCH AND CHANGES TO FDA DRUG APPROVAL PROCESS

On December 13, 2016, President Barack Obama signed the 21st Century Cures Act into law after it was passed by both the House of Representatives and the Senate by nearly unanimous votes.

The legislation covers many healthcare topics. Two are of particular importance. The first expands funding for the National Institutes of Health by \$4.8 billion over 10 years, or about 1.3 percent a year. The additional funding is directed to the Precision Medicine Initiative, the Cancer Moonshot Initiative, and research on the brain and brain disorders. The funds will have to be appropriated annually.

The second topic involves the approval process for new drugs by the Food and Drug Administration (FDA). Several sections address greater patient involvement in drug approvals and advancing new therapies to address unmet medical needs with respect to rare disorders or conditions that are serious or life threatening. One section addresses accelerated approval for regenerative therapies, including cell therapy, therapeutic tissue engineering, and human cell and tissue products.

These changes will not take place overnight. The legislation provides timelines for the FDA to develop initial guidance, followed by public consultation. These will be multiyear processes which the FSH Society will closely follow.

— Morgan Downey
Washington, DC, Counsel



8 TELESCOPING MAGNETIC PICKUP TOOL

A very handy device for picking up small- to medium-size metal objects like cutlery, keys, or pens. These sell for about \$6 on Amazon—and Harbor Freight Tools sells comparable models for slightly less. Are you catching a “clumsy vibe” here?

9 TELESCOPING WALKING STICKS

I bought a pair on Amazon for about \$26 and use them both in tandem like ski poles, as well as using individual sticks along with my normal cane. They can be adjusted from about 26 to 53 inches in length, so they can fit into luggage without a problem.

In addition to these helpful devices, I've learned a few tips and tricks that make a big difference. I hope these tips and products can help you as they have helped me—and if you know of any others, please share them with the FSHD community!

- 1 A nod to our son, Sam, who programmed the driver's seat memory in my car—using the second setting to raise the seat height to the max and tilt all the way forward. It adds a little more than two inches in height from my normal driving position.... Every little bit helps! I use this second memory position to help me get into and out of my car with less discomfort, putting the seat into the first memory position once I'm inside and ready to drive.
- 2 If you have difficulty getting up from a seated position, have a helper grab a belt loop on the back of your pants and gently pull upward as you raise yourself out of your chair with your arms. It doesn't take much extra boost to do the trick.
- 3 If you think you need a new mattress soon to help you sleep comfortably, try a memory foam mattress topper as an interim step. We found that it made a huge difference (for both of us) and four years later are still using the same one.
- 4 This isn't really FSHD specific, but many of us are at the age when hearing aids are in our futures (or already a part of our daily lives). I had known that aids were in my future for some time, but had balked at the high cost. Then I checked out Costco's pricing and took the plunge. Their store-brand Kirkland devices were about \$1,800 for a pair, much less expensive than big-name brands, and they are manufactured by a well-known leader in the industry. The quality is excellent, and the nearly invisibly tiny units have been a lifesaver.

My Boston adventure

Traveling solo with my wheelchair

by **TRISHA LYNN SPRAYBERRY**
Las Vegas, Nevada

My adventure began late Wednesday night, November 9, 2016. My fiancé, Erin, had dropped me off at the airport. With my suitcase in hand and a kiss to send me on my way, I was headed to Boston on a red-eye flight. I was attending my first patient conference, hosted by the FSH Society, and their inaugural #CureFSHD gala. It was at the grand Westin Copley Place Hotel in Boston, Massachusetts.

What makes this trip special for me is the fact that this was a solo adventure. Traveling alone is no big deal for most people, but for me, since I have facioscapulohumeral muscular dystrophy and am dependent on the use of a wheelchair, traveling alone to Boston was an anxiety-ridden challenge. A test, if you will, one that I was more than willing to not only face, but to obliterate.

This was more than just a trip to Boston. This was a proving ground, and I had to succeed. Not because I want to show the world the capabilities and true independent power of those with disabilities, not even to demonstrate the true strength one can have with FSHD. I had to do this to prove to myself that if I can find a way to accomplish this journey alone, then I can find a way to accomplish anything.

I know it sounds like a cliché to say that, but it is true. This really was a magical and transformative experience, but even now, I fail to find an adequate way to describe the emotions that it evoked.

Once I had parted from Erin, I went straight to the ticket counter. I checked my suitcase and was escorted through TSA and to my gate. By 9:50 p.m., I was separated from my power wheelchair and transferred into the airline's aisle chair. When you are being strapped into the airline's wheelchair, with the two shoulder straps and the lap belt, and then pushed backwards onto the plane and down the aisle to your assigned seat, it leaves you feeling like Hannibal Lecter, sans the mouth guard and reputation, of course.

I settled into my seat, the rest of the passengers boarded, and off we went for



“One moment of the night that stuck out was speaking to a researcher about why we were all here. She had said that for her, it really brings home the work that she does and puts the face and heart and strength in what she does. She had the most beautiful, humbled, real smile on her face. She said that when she is in the lab, it's all business, but when she came to this conference, it gave her work its true home. I had told her that for me, being a patient, coming to the conference does feel like home. Out there in the real world, FSHD is a rare disease; it's virtually unheard of. And no one cares. But here at the conference, these are all people who care, who are also just like us, whereas out there, we may not know another with FSHD unless it's family. And you can feel the love and the dedication and the heart of why we are all here.”

—TRISHA SPRAYBERRY

a smooth flight, landing at Boston's Logan Airport about 6:30 a.m. local time, Thursday morning. After de-boarding the plane in the same fashion, I was wheeled by an airport attendant to claim my wheelchair and luggage at the baggage claim.

The attendant kindly offered to call me a taxi, which I accepted, and then accompanied me to my ride. The second I reached the doors to exit the airport, my body tensed and seized my breath. It was freezing outside. I didn't think much of the weather, though. I was just so relieved that my wheelchair and I both made it through the flight safely in one, functioning piece. That part of this whole trip was the thing that I had been the most worried about.

I arrived at my hotel and was checked into my room by 8 a.m. A huge thank you is owed to the FSH Society and SHIFT Communications for securing my accommodations for this trip. They had worked very hard the week before the conference on finding an accessible room for me, when ADA rooms at the conference hotel had sold out. Without their generous efforts, I never would have been able to have this experience.

After my look around the hotel room, I spoke with my family and went to sleep. After a long nap, I spent the evening unpacking, talking with my family, eating dinner, charging up my wheelchair battery, and connecting with fellow FSHer and dear personal friend, Susie, making plans to meet up the next day at the conference.

Friday morning went by like a breeze. I awoke refreshed and ready for the excitement of the day. The concierge called a taxi for me, which arrived within 15 minutes. The personal service of this hotel was above and beyond. Thank you, DoubleTree, for taking such good care of your guests!

When I arrived at the Westin, the entrance was crowded with bodies of people coming and going. Some of the bodies drew my attention. They were different from the others, and this made me smile. They were different in the same ways that I am, because they also have FSHD. When I

saw those bodies, it was my body as it is, or as it was at an earlier point in my life. I had found a little piece of myself connected in this way to this world.

I followed the others inside and to the elevator, and the strangers waiting for the lift with me felt as familiar to me as family. This only made my eagerness grow. The two-floor ride felt like waiting to reach the top floor of the Empire State Building. I had grown so nervous, but I don't think I was the only one. It was a very quiet ride up to the third floor.

I spotted my friend Susie outside the conference hall where the Patient Connect Conference was being held. She introduced me to her companion, her loving and supportive son. After touching base with them, I wandered around to find a bite to eat before things started up with the conference. It was worth the time to tour the beautiful architecture of the Westin Hotel.

I rolled my way back into the conference and found the table where Susie and her son were sitting. On my way over, I ran into Dan Perez, one of the co-founders of the FSH Society. I'll admit, I felt a little bit star struck shaking his hand when we met, as he was zooming off to turn down the room lights for the presentations the researchers were giving.

I immediately was absorbed into the presentations. These researchers are on the frontlines of FSHD. These people are tirelessly dedicated to finding a cure for those of us affected. For me. And anything they had to say, I needed to hear.

I wasn't the only one who thought this. Almost everyone in the room had either their pencils frantically scribbling notes down on their itineraries or napkins, or they had their phones poised to snap shots of the slides projected at one of the projection screens raised on both sides of the speaker's podium.

There were rounds of questions and comments from patients, family members, and other supporters of FSHD research, and the researchers, doctors, and members of the FSH Society provided great insight into the breakthroughs our FSHD warriors are making toward treatments and possible cure

avenues, such as CRISPR, a gene-editing tool. Many also contributed intimate stories and perspectives into the world of those living with FSHD.

During the break, everyone had a chance to mingle. This is when I had the pleasure of meeting quite a few people who over the years had become so familiar to me through our FSHD community on social media. Carden Wyckoff was there. She and her brother just completed a piggyback trek in the Appalachian Mountains. Carden is quite charming in person, her smile infectious.

ride away, I should have the time to go back to my hotel and change into more suitable attire for the evening gala. Well, I was wrong. The taxi was late, and by the time it arrived, I was being handed my name badge for the gala.

The gala began with cocktails with the "rock stars of FSHD research." Guests arrived in classy evening wear, accessorized with a passion for a cure. The wait staff went around the room offering morsels of coconut chicken, macaroni and cheese bites, and bacon-wrapped scallops, while



“Positive energy exuded from the 200-person crowd as the hum of small talk grew into a cloud of conversation engulfing the room. It felt electric, and, it felt like a blow to the chest, humbling me. I found myself holding my breath in awe.”

—TRISHA SPRAYBERRY

I also got to meet for the first time many whom I have grown to admire and respect, leaders certainly in the eyes of our community: various doctors, researchers, donors, and members of the FSH Society and other organizations. Sharing a hug with June Kinoshita for the first time was definitely my favorite moment.

I took advantage of the intermission to call a taxi ahead of time. I had decided before I left my hotel that morning that since the Westin was only a 10-minute taxi

the bar offered spirits tailored for the evening. The "Red Cure" was a delicious cranberry concoction.

Positive energy exuded from the 200-person crowd as the hum of small talk grew into a cloud of conversation engulfing the room. It felt electric, and, it felt like a blow to the chest, humbling me. I found myself holding my breath in awe.

We were all swept into dinner, a wave of bodies filing into the same hall where

... continued on page 19

A tunable FSHD-like DUX4 mouse model

Now available through The Jackson Laboratory

by PETER JONES, PhD, and TAKAKO JONES, PhD
University of Nevada, Reno

The aberrant expression of the DUX4 primate retrotransposon is the key mediator of all forms of FSHD. Thus, the DUX4-fl mRNA and protein are prime targets for therapeutic intervention.

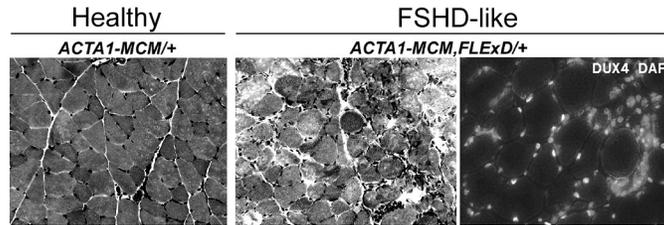
Our laboratory at the University of Nevada, Reno School of Medicine reports the successful generation and free distribution of a viable, fertile, and highly tunable phenotypic FSHD-like transgenic mouse model based on the controlled expression of DUX4. This mouse, referred to as FLExDUX4 (or FLExD), is now available from The Jackson Laboratory as B6(Cg)-Gt(ROSA)26Sortm1.1(DUX4*)/Plj/J, catalog #028710 (<https://www.jax.org/strain/028710>).

FLExD mice are available as males and females, hemizygous and homozygous, and will be ready for shipping after February, 2017. These mice are all very fertile, and have normal lifespans, and produce large litters.

DUX4 expression can be induced through expression of the Cre recombinase by a number of mechanisms but typically by mating with Cre-expressing lines of mice, resulting in an FSHD-like pathology.

The flexible design of this mouse model allows the investigator to induce DUX4 expression in either young or adult animals as well as control the degree of pathology and rate of disease progression, thus allowing for the assessment of prevention, inhibition, or reversal of pathology as desired, dependent upon the therapeutic intervention being tested. In addition, one can distinguish early, initiating events of pathology from cumulative, chronic pathological effects. The FLExD mouse can essentially be tailored to the type of therapeutic being tested or pathway being studied.

It is widely recognized that mouse-based preclinical studies have often produced inconsistent or even incorrect results due to lack of experimental rigor and/or lack of transparency in methodology, ultimately leading to poor reproducibility among labs



H&E staining shows severe muscle pathology in gastrocnemius muscles of ACTA1-MCM, FLExD severe FSHD-like model mice (center) compared with ACTA1-MCM healthy control mice (left). DUX4 immunostaining (right) shows mosaic expression and the nuclear localization of DUX4 protein in gastrocnemius muscle after TMX induction. Figure courtesy of Jones laboratory.

and failures of clinical trials. The NIH has instituted a new policy demanding that investigators address the critical issues of rigor, transparency, and pertinent biological variables. We fully support this initiative and believe that a poorly characterized and inconsistent FSHD-like mouse model would only serve to set the field back, wasting time, money, and resources. Therefore, we have spent nearly two years characterizing the FLExD model and verifying aspects of its phenotype in collaboration with experts in several other labs.

Removing barriers, particularly the restrictions institutions place upon usage of lab-generated and patient-derived biomaterials, is critical to accelerating therapeutic development in FSHD. The ethical obligation to deposit cell and animal resources into publicly accessible biorepositories for unfettered access is critically important for the entire field. Thus, we have balanced our desire for a complete characterization and natural history study (which is ongoing) with the level of characterization required to make this model a usable tool for the FSHD field. Importantly, our model is highly consistent and reproducible within animals, between animals, and over many generations.

Therefore, in an effort to facilitate fast and clean transfer of animals and to relieve restrictions or complications imposed by investigator control of reagents and spur FSHD therapeutic development and preclinical testing, the currently unpublished FLExD line of inducible FSHD-like mice is now freely available from JAX labs.

Key elements of the FLExD model:

- Viable and fertile as FLExD/+ and FLExD/FLExD
- FLExD mice live a normal lifespan (>2 years)
- Very low level of DUX4-fl mRNA and protein in the absence of Cre
- Viable and healthy double transgenic animals when mated with ACTA1-MerCreMer tamoxifen-inducible line
- Chronic low levels of DUX4-fl expressed throughout life with investigator-controlled bursts of DUX4-fl, recapitulating how we envision DUX4 expression in FSHD patients
- No adverse effects on transgene expression or inducibility through >6 generations
- DUX4-fl mRNA and protein expression is consistent throughout an animal's musculature (contralateral muscles can be used as controls)
- FLExD x ACTA1-MCM cross produces large litters (10-12 pups) with many double transgenic animals
- Five severity levels of FSHD-like phenotypes have been analyzed
- Cre induction of DUX4 expression produces consistent pathophysiology from animal to animal
- Histopathology is consistent within each severity model
- Several other Cre expressing mouse lines of potential interest have been tested

Specific details for achieving the various severity models will be included in a publication sometime within the next year. Until that time, many details are available on our lab website, <http://med.unr.edu/jones-lab>. Investigators can also contact Peter Jones (peterjones@med.unr.edu) or Takako Jones (takakojones@med.unr.edu) at the University of Nevada, Reno School of Medicine, and we will be happy to help out.

This work was funded by the FSH Society (FSHS-22012-01), the Chris Carrino Foundation for FSHD, MDA (#383364), and N IH/NIAMS (R01AR070432, R21AR070438). 



FSH Society 2017 Calendar

New meetings are being added all the time. Check the website for updates: <https://www.fshsociety.org/fsh-society-events/>. Don't see your city on the list? Contact robyn.oleary@fshsociety.org to find out how to start your own patient meeting.

Saturday, March 4, 2017
11:00 a.m.–2:00 p.m.
San Diego FSHD Support Group Meeting
Leary Medical Institute Building
San Diego, CA

Tuesday, March 7, 2017
5:00 p.m.–7:00 p.m.
FSH Society New England meeting
Lexington, MA

Saturday, March 11, 2017
3:00 p.m.–6:00 p.m.
FSH Society New York City meeting
West 89th Street
New York, NY

April 22, 2017
FSH Society Dallas meeting
Irving, TX

Sunday, April 23, 2017
2:30–5:30 p.m.
FSH Society Austin meeting
Austin, TX

Saturday, April 29, 2017
FSHD Physical Therapy free screening
re+active Physical Therapy & Wellness
Los Angeles, CA

Sunday, May 7, 2017
11:00 a.m.–3:00 p.m.
Miami FSH Society meeting
Shake-a-leg, Miami, FL



The 4th Saturday of Every Month
10:00 a.m.–12:00 p.m.
Western Washington FSH
Community Skype



Saturday, May 20, 2017
Minnesota FSH Society meeting
Location TBD

Saturday, June 10, 2017
8:00 a.m.–10:00 a.m.
Cosie Laurrello Memorial Run & Walk
Fischer's, Pine Lake
Jefferson, OH

Sunday, June 11, 2017
FSHD Family Day Conference
Nationwide Children's Hospital,
Columbus, Ohio

Thursday, June 15, 2017
Team FSHD Cycling Kickoff Party
Urge Gastropub
Oceanside, CA

Saturday, July 15, 2017
FSH Society Family Day conference
(afternoon) & 4th annual Songs in the
Key of Steven Blier (evening)
San Francisco, CA

Saturday, September 9, 2017
9:00 a.m.
Walk & Roll to Cure FSHD 2017
Phillip S. Miller Park
Castle Rock, CO

Saturday, September 16, 2017
6:00 p.m.–10:00 p.m.
Musclepalooza 2017
Hope Ridge Farm
New Hope, PA

October, 2017
Date TBD
FSH Society Family Day conference and
3rd annual Ghostly Gala to Vanish FSHD
Los Angeles, CA

Ways to engage

SIGN UP FOR EMAIL NEWS!

Get our email alerts for the latest news, event notices, clinical trial info, and more. Sign up on our website by clicking "JOIN". If you are certain you are on our email list, please check your spam or junk folder.



GET SOCIAL!

Find our Facebook, Twitter and Yahoo! Groups by visiting www.fshsociety.org and clicking on the logos in the right-hand margin. Our online communities are great sources of news, advice, and social support. The FSH Society



Yahoo! Groups forum, online since the 1990s, has tens of thousands of searchable posts. Bookmark these pages and come back often. Use your account privacy settings to limit who can see your posts.

HAVE YOU MADE A GIFT TO THE SOCIETY IN 2017?

Thanks to the support from members like you, the FSH Society is a world leader in combating muscular dystrophy. Your donations are tax deductible, and they make a real difference. Please send your gift in the enclosed envelope. Or contribute online at www.fshsociety.org. Thank you!



COMMIT TO THE FUTURE

Consider taking your gift to the next level by making a planned gift. A bequest or endowed fund helps to ensure that the FSH Society has the long-term support to sustain its mission. Questions? Please contact June Kinoshita at (781) 301-6649 or june.kinoshita@fshsociety.org.

Always check with your advisors when making a change in your will or estate plans, and learn how current tax laws and other legislation may affect your plans.



Ask the physical therapist (part 2)

Julie Hershberg answers your questions

The following is Part 2 of the transcript of a question-and-answer session conducted over the FSH Society's Facebook page, with Julie Hershberg, PT, DPT, NCS. Hershberg is a physical therapist who is a Board Certified neurologic specialist. She practices at [re+active] physical therapy & wellness and is an instructor in the Doctor of Physical Therapy program at the University of Southern California.

Q. What's the best exercise for arms unable to move above shoulder height?

A. Oh, yes—a very big question! Again, this depends on how much movement and strength you have, so I cannot give specific advice, but I will offer these general suggestions.

I often recommend exercises in which you do not have to move against gravity. A good example is a shoulder exercise lifting the arm to the side while lying down. This provides good scapular support, and you don't have to lift against the force of gravity. I also love using supports such as mobile arm supports, TRX, or even new technology such as Redcord (I haven't used this, and I don't have it, but I have seen it used in other facilities): <http://www.redcord.com/Portal.aspx?m=546>. In the TRX or Redcord, you can take up the weight of the arm in order to perform more exercises in the ideal alignment or posture.

One thing to consider is that, depending on the degree of weakness of a person with FSHD, some muscles may be working or exercising to their maximum just to perform everyday activities. A weak scapular muscle such as the serratus anterior or middle trapezius, for example, will be challenged to complete daily showering, hair washing, and hair combing tasks. These specific muscles may need to rest and not perform additional resistive exercises.

I don't think that PT is actually necessary forever—maybe for a short time to get you started on a good program, then getting started with a therapist and transitioning to work on your own or at a gym. The CDC recommendations for exercise are for a total of 150 minutes a week for healthy living!

Q. With wasting occurring asymmetrically (right/dominant arm faster than



A physical therapist positions a patient so that her arm can go through a wider range of motion. (Photo courtesy of K. Eichinger.)

left), how should I modify my exercise regimen? Should I consider wearing some kind of undergarment support, or is it better not to give the muscles this support and make them work?

A. First, for your exercise routine—you should consider exercising each limb at its own capacity—for example, not lifting the same amount of weight in both arms. This will be very specific to your individual strength. I am not sure which type of undergarment support you are referring to, but I will recommend something like an abdominal binder for people who have very weak abdominals because it will help prevent overuse of other muscles for postural support and can help decrease some postural deformity that can occur over time. Wearing a support does not necessarily mean that you don't use the muscles (in fact, sometimes they work better), and most people will report improved function.

Other compression garments for training (for example, the compression socks or arm sleeves) could also potentially be helpful (I certainly don't think they would be harmful unless you have a circulatory disorder, so please consider your other health history if that is relevant). While there is not definitive research on these types of garments and performance, my philosophy is: If you feel better and are more likely to exercise and do the things you love with it, then use it!

Q. What is your thought on canes/braces for weak legs? Will they cause dependency?

A. My first advice is for people to use devices to keep them independent and enjoying life as much as possible. If wearing a brace or using a cane or wheelchair lets you do the things you love, then absolutely—the benefits outweigh the risks, and your quality of life will soar.

However, there are some things to consider: First, for bracing—there is not evidence to show that wearing a brace decreases muscle activity. This is a common myth. Most people will be able to walk faster and more smoothly with the brace on, and therefore feel more comfortable wearing it.

The same is true for a walker or cane. Devices for walking can immediately make you feel more comfortable and confident. One thing I often recommend is trying the devices and seeing if you walk farther or have fewer trips and falls—and then, in that case, you will walk more and end up stronger and more fit in the long run.

My advice is to use the least restrictive device that gives you the most freedom, safety, and independence. A PT can usually do trials with all kinds of devices to help you choose the best one for you.

Q. What is the best exercise for a weak leg causing strain in the calf muscle?

A. This is a tough one. There are many reasons why the calf can be strained, so this requires assessment by a PT. For example, calf strain can be due to hip weakness (and therefore overuse of the calf muscle to propel

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Team FSHD Cycling eyes epic race in June 2017

3,000-mile race to raise awareness and funds

by JUNE KINOSHITA
FSH Society

During the month of June of this year, the FSHD community will enjoy the thrill of having one of its own lead an eight-person team in the Race Across America (RAAM), a bicycle race from Oceanside, California, to Annapolis, Maryland. The team, recruited by George Pollock, plans to compete in the Open Relay Division, and expects to complete the race in under seven days and cover more than 450 miles a day.

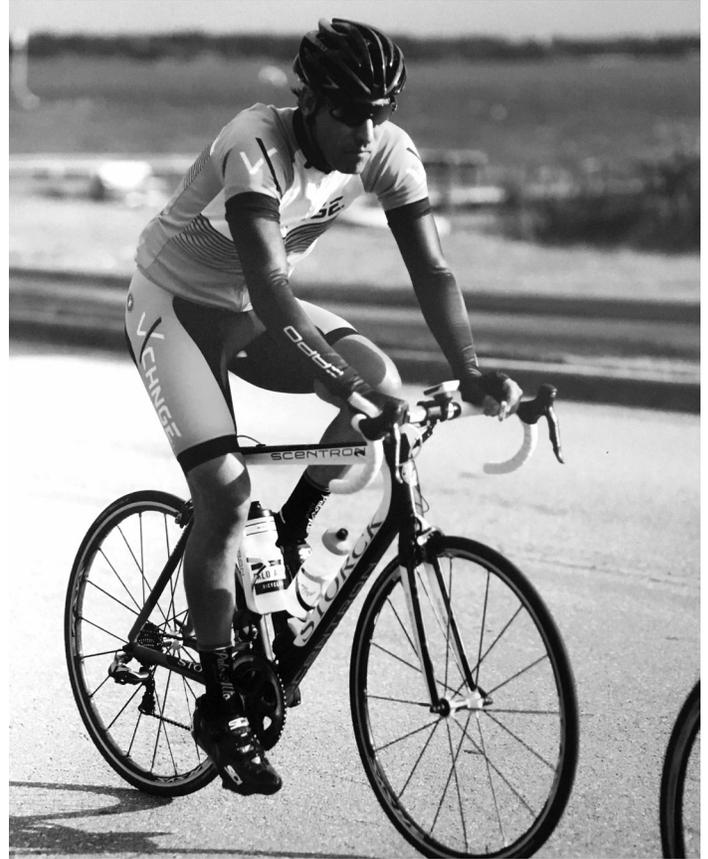
The team has a personal connection to the disease. Pollock has FSHD. He started to have muscle weakness in his lower right leg while in his late twenties. Ten years later, when he experienced muscle weakness in his left leg, he was finally diagnosed with FSHD. Fortunately for him, the disease is progressing slowly. He was recently elected to the Board of Directors of the FSH Society and has made a commitment to promote awareness to improve diagnosis, support patients, and raise money to fund the Society's mission.

The Race Across America (www.raceacrossamerica.org) is one of the most respected and longest running sports events in the world. RAAM has a rich and storied history dating back to four individuals who raced from Santa Barbara to New York City in 1982. In 1992, relay teams were added and became the most popular and fastest growing segment of the race. In 2017, RAAM will celebrate 36 years of racing.

RAAM has had riders from five continents and 35 countries. In the last 10 years, the race has been used to promote awareness and fundraising for dozens of charities, and racers have raised more than \$10 million. The race attracts significant media coverage around the world and can be followed live at www.ridefarther.com.

"There is no other race like it in the world," said Pollock. "Our team is looking forward to participating in such an iconic event, and raising awareness for FSHD and the efforts to find a cure."

Team FSHD Cycling has set a goal of raising \$101,000 for the FSH Society, net of all expenses. Others are encouraged to support the epic effort. FSH Society Board member Amy Bekier has already taken up the banner and is planning a dinner in Oceanside, California, where the race starts, to celebrate Team FSHD Cycling and raise additional funds. 



George Pollock shows off his racing form.

Please visit the FSH Society website for updates, and visit the Team FSHD Cycling website (www.fshdcycling.com) to find rider and race information and to support Team FSHD Cycling.

Note: Team FSHD Cycling is holding its 2017 Race Across America Campaign as independent volunteers, not as employees, agents, Board members, advisors, or volunteers of the FSH Society.

ASK THE PHYSICAL THERAPIST (PART 2)

... from page 14

the leg forward). Calf strain can also be due to calf weakness or tightness as well. Again, I recommend an individualized evaluation to help discern the best place to start.

Q. What is the best exercise for weak stomach muscles?

A. Alas—one more time where I will say that this really requires a thorough

assessment of your strengths. In general, I recommend strengthening of the deeper abdominal muscles, because these are key to stability and decreasing pain (and can often be intact when some of the larger muscles are weakened in FSHD).

I like this PT video about how to contract the deep abdominal muscles as a starting point (there are many variations and progressions to this exercise): <https://www.youtube.com/watch?v=p8O04WLXFt8>.

In general, PTs are very good at identifying the control you have, and then progressing and strengthening it in various postures and positions. I have also worked with Pilates instructors and trainers who have good applications of these methods.

Another thing to consider is using an abdominal binder to help support the abdominal muscles and put you in a better alignment for using them in function. 



2016 CureFSHD Gala

FSH Society honors leaders in science and advocacy

by JUNE KINOSHITA
FSH Society

On November 11, the FSH Society honored four distinguished researchers and community members at its inaugural CureFSHD National Gala in Boston, Massachusetts. The event, hosted by WBUR's *Morning Edition* Host Bob Oakes, celebrated 25 years of progress toward finding a cure for facioscapulohumeral muscular dystrophy (FSHD). The gala, which attracted more than 250 researchers, scientists, FSHD patients, and supporters, was considered a great success by attendees and organizers alike.



Daniel Perez (right) presented Professor David Housman (center) of the Massachusetts Institute of Technology with the 2016 FSH Society Service Award for his seminal role as chair of the Society's Scientific Advisory Board.

“Our inaugural National Gala was a chance for the entire FSHD community to come together to recognize exceptional individuals who have dedicated their lives and careers toward advancing FSHD research,” said Daniel Perez, co-founder, president, and CEO of the Lexington-based nonprofit FSH Society. “While there are many worthy candidates, this year we recognized a research pioneer whose work has played a major role in establishing the scientific foundations of the FSHD field and a young investigator who has published high-impact work and shows exceptional promise to develop into a leader. We also honored two pivotal members of the FSH Society community for their commitment to building the organization.”

The 2016 FSH Society Pioneer Award was received by Silvère M. van der Maarel, PhD, who has made seminal contributions to the identification of the unifying genetic mechanism underlying FSHD. Professor van der Maarel was trained as a human geneticist at the Radboud University Nijmegen Medical Center in the Netherlands. His scientific interests focus on the genetic and epigenetic bases of FSHD.

The 2016 FSH Society Young Investigator Award was given to Charis L. Himeda, PhD, for many innovative studies, including her contributions to a ground-breaking 2015 paper published in

Molecular Therapy. Dr. Himeda, an Instructor in cell and developmental biology in the lab of Peter Jones at the University of Massachusetts Medical School, published a widely noted study demonstrating the use of CRISPR gene-editing technology to suppress a gene that plays a key role in FSHD.

The CureFSHD National Gala was also a chance for the FSH Society to recognize two extraordinary individuals who have provided many years of dedicated service to the FSH Society.

The Society's Service Award was presented to David E. Housman, PhD, Ludwig Professor of Biology at Massachusetts Institute of Technology. A renowned scientist and teacher who has been an inspiring mentor to hundreds of scientists for nearly half a century, Housman has raised awareness of FSHD by including it in the MD/PhD curriculum of Harvard-MIT Division of Health, Sciences and Technology. Under his leadership as chair of the FSH Society's Scientific Advisory Board, the SAB has reviewed hundreds of grant applications, resulting in \$7 million in grants awarded to date and more than 300 publications in top-tier journals.

The second FSH Society Service Award was given to Chris Stenmon, a member of the FSH Society Board of Directors from 2006 to 2014, who served on the Finance and Development committees as well as being treasurer for 2014. Stenmon was diagnosed with FSHD as a teenager and began his involvement with the Society by organizing and hosting a fundraiser beginning in 1998 which evolved into a major annual event raising nearly \$300,000 over the years.

“The FSH Society has been there for me whenever I needed them,” said Stenmon. The founders, “with their perseverance, drive, and will to never give up, are the main reasons we have made so much progress toward treatments and a cure.” FSH Watch

RAZOO ONLINE FUNDRAISING

Razoo makes it easy to create an online campaign. Your donors will enjoy the convenience, knowing that their gifts will go directly to the FSH Society. With Razoo you can easily promote your campaign over Facebook, Twitter, and other social media. <http://www.razoo.com/story/Facioscapulohumeral-Society>



DONATE YOUR EBAY SALES EARNINGS

The FSH Society is registered (as “FSH Muscular Dystrophy Society”) on eBay's charity auction site. If you have an eBay seller's account, you can list items and direct from 10 to 100 percent of the proceeds to the Society.



FREE MONEY!

Many organizations will match employees' charitable donations, or will donate if employees volunteer their time to a charity. Don't leave this money on the table! Ask your employer about their charitable gift programs.

GIVE WITH A SMILE THROUGH AMAZON

Amazon will donate 0.5 percent from your eligible purchases to the FSH Society whenever you shop on AmazonSmile. To get started, register here: <http://smile.amazon.com/ch/52-1762747>.



A second promising FSHD mouse model

Expresses DUX4 only in muscle

by **SCOTT HARPER, PhD**
Nationwide Children's Hospital, Columbus, Ohio

Mouse models of disease are important tools for developing therapies. During the past decade or so, several attempts have been made to generate FSHD mouse models that express the DUX4 gene in their chromosomes.

Although these models were designed logically, the animals were difficult to produce, and they did not show the muscle weakness and damage seen in humans. These first models also suggested that it was difficult to make mice expressing human DUX4, because the gene was toxic and incompatible with normal mouse development.

We concluded that if we wanted to make a DUX4 mouse, we would have to tightly control when and where it could be turned “on,” and began working to generate a new FSHD mouse model in 2009. After many difficulties, we finally successfully produced a model in which DUX4 could be turned on only in muscles.

Our new mice are able to reproduce easily, express DUX4 in muscle, and develop features of muscle disease in the whole animal and microscopically. For example, compared to normal mice, our DUX4 mice have weaker muscles, microscopic evidence of muscle cell damage, are less active, and have an unsteady and slower gait.

We are using these animals to test therapies for FSHD targeting DUX4, and are making them available to any researchers in the FSHD field who think they may be useful. We would like to thank the Muscular Dystrophy Association, FSH Society, and their donors and scientific boards for supporting this work.

Technical summary

These mice contain a CRE-inducible transgene knocked into the ROSA26 locus as a single copy. A FLOXED-Neomycin resistance gene is cloned upstream of a V5 epitope-tagged DUX4. The construct also has the 3' UTR including the pLAM poly A. We breed the mice to the HSA promoter-CRE-ERT2 line available from Jackson Laboratory.



Members of the Harper Lab, from left to right: Mandi Speakman, Diana Mukweyi, Nizar Saad, Jocelyn Eidahl, Scott Harper, Allison Fowler, Mustafa Alkharsan, Nettie Pyne, Lindsay Wallace, Carlee Giesige.

Double transgenics are viable, have zero DUX4 leakiness, and we get Mendelian ratios. DUX4 can be turned on with Tamoxifen. At high doses of Tamoxifen, animals are non-ambulant by seven days. At low doses, animals show a gait defect, muscle histopathology, TA-specific force deficits, and significantly reduced activ-

ity. (Note: the mouse genes are artificially engineered so that the drug Tamoxifen can be used to switch DUX4 on. Normally, Tamoxifen had no effect on DUX4.)

For questions and to request mice, please contact Scott Harper. Phone: (614) 355-2893. Email: scott.harper@nationwide-childrens.org. 

YOUNG HOLLYWOOD STEPS UP FOR A GHOSTLY GALA

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Yura—created a spectacular event, with many returnees from last year's lineup: renowned comedienne Wendy Liebman as Mistress of Ceremonies, Orchestra Leader Dean Mora and His Band, and Live Auctioneer Zack Krone.

An entertaining and innovative digitally animated invitation was created and donated by award-winning animation director Barry Jackson, with a soundtrack mashup contributed by NYC-based Marshall Weinstein of SET Artist Management.

Genetic research scientist Amanda Rickard returned for a report of the year's advancements in clinical trials and progress toward finding a cure for FSH mus-

cular dystrophy. Sponsors aTyr Pharma and the Abundance Foundation generously came on board once again.

A mere three weeks before show time, FSHD champion Max Adler (of *Glee* and *Sully* fame) presented a daunting but spectacular opportunity: Entertainment industry heavyweights Maria Calabrese and Ronnie Ward of Griff Entertainment proposed to donate \$15,000 in services to create a livestream platform, and to produce and direct a broadcast of A Ghostly Gala.

All that was needed was a matching \$15,000 to cover camera and audio equip-

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locus on chromosome 4. In FSHD1, this is caused by a shortening of the D4Z4 array of repeated units at the disease locus. In FSHD2, loss of repression is caused by mutations in SMCHD1 and other proteins that normally maintain silencing at D4Z4 arrays.

In both situations, the failure of normal repressive mechanisms causes the molecular structure of the FSHD locus to relax, allowing abnormal expression of the DUX4 gene from the distal end of the array. The DUX4 protein then activates a host of other genes, ultimately leading to pathology in skeletal muscle. Thus, there are two viable therapeutic avenues for blocking expression of DUX4: promoting repression or inhibiting activation of the FSHD locus. Several labs and companies are actively screening compounds with these aims in mind.

Although the underlying cause of FSHD is now well established, the mechanisms by which the DUX4 protein causes muscle weakness and degeneration are still unclear, although a number of cellular processes have been implicated. While overexpression of DUX4 is highly toxic to muscle cells, lower or more sporadic expression—perhaps more relevant to the situation in FSHD patients—leads to subtler defects in RNA and protein metabolism, maturation of muscle cells, and immune functions.

Likewise, the role of DUX4 toxicity during development, from embryo to adult, remains to be understood. When is DUX4 expressed over the course of a patient's

lifetime—early on in the womb, with no apparent consequence, or suddenly during childhood or adulthood, with immediate consequence? Is DUX4 expressed in muscle stem cells, and if so, does it contribute to the inability of FSHD muscle to regenerate?

Fortunately, a growing number of animal models—some of which mimic key aspects of the disease—are being developed and characterized to answer these questions and provide an *in vivo* platform for therapeutic testing. In an exciting development, one of these models, an FSHD-like mouse that expresses controllable levels of DUX4, is now available to all investigators from The Jackson Laboratory (see story on page 12).

Just as mutations in different genes can often lead to the same disease, different mutations in the same gene can also result in different diseases. A striking example of this phenomenon was reported for arhinia, a rare developmental disorder characterized by the complete absence of an external nose, often with eye or reproductive defects. Arhinia is caused by mutations in SMCHD1, a protein that represses repetitive elements and the most commonly mutated gene in FSHD2.

While SMCHD1 mutations in FSHD2 patients are scattered across the entire gene, mutations in arhinia patients are tightly clustered around a specific region of the gene. Interestingly, examination of the FSHD locus in arhinia patients revealed the marked loss of DNA methylation characteristic of FSHD2. It is unknown whether

these arhinia patients would go on to develop FSHD symptoms later in life, but these results indicate that other functions of SMCHD1—beyond its role in establishing repressive DNA methylation—play a key role in determining disease outcomes.

Studies such as those in arhinia—and presentations by other newcomers to the field—are a testament to the growing interest in FSHD from clinicians and experts in diverse areas. This interest was matched by a record number of industry sponsors and partnerships, as biotech and pharma companies are now deeply engaged in the identification of therapeutic targets for drug development.

This flurry of productivity comes on a wave of progress in the development of promising new therapeutic avenues. The first use of CRISPR technology in patients—gene editing as immunotherapy for an aggressive form of lung cancer—was recently attempted by a group in China, and the first CRISPR clinical trials in the US—also for cancer immunotherapy—are rapidly approaching.

While FSHD faces unique therapeutic challenges, the field can only benefit from progress made in the treatment of other disorders and lessons learned by others on the road to a cure. FSH Watch

Editor's note: Charis Himeda, PhD, is a research assistant professor in the Department of Pharmacology at the University of Nevada, Reno School of Medicine.

YOUNG HOLLYWOOD STEPS UP FOR A GHOSTLY GALA



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ment and crew for the day of the Gala. Within 72 hours, Todd Turner of Junket Productions (creators of international press junkets for large-scale studio films) came through with a generous donation of everything needed.

The team then created a website to promote the livestream, and in a stunning show of friendship and support for Max Adler, young Hollywood came out for the FSH Society. Mayim Bialik (*The Big Bang Theory*), Ryann Redmond (*If/Then; Bring It On*), Jayson Blair (*Whiplash*), Larisa Oleynick (*The Secret World of Alex Mack*), Katie Leclerc (*Switched at Birth*), Stephen Kramer Glickman (*Storks*), Josh Sussman

(*Glee*), Josh Kelly (*UnREAL*), Chester Rushing (*Stranger Things*), and Kayla Ewell (*The Vampire Diaries*) contributed video selfies which were then cut together by Ellen Rennell into an inspiring invitation to promote the livestream and raise awareness of FSH muscular dystrophy.

Public awareness of FSH muscular dystrophy made a giant stride forward when, in response to the celebrity selfie-video, *Peoplemagazine.com* picked up the story of the livestream and tweeted it to seven million followers. The next day, Hollywood blogger Perez Hilton retweeted it to an additional six million people. The livestream exploded into a three-camera,

global broadcast complete with live interviews and interstitial “infomercials” about FSHD pulled from the Society's archives.

Precisely at noon on Sunday, October 30, 2016, a team of more than 30 determined and impassioned volunteers descended to join the Cicada Club staff and FSH Society members and staff to prepare for the event. At 5 p.m., arriving guests were greeted on the red carpet by a backdrop of a huge bust of Johann Sebastian Bach made up as Ziggy Stardust.

The “Music Through the Ages” theme had obviously been taken to heart! In attendance were Beethoven, Jerry Garcia,

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MY BOSTON ADVENTURE

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the patient conference was held earlier in the day, the space now transformed into a breathtaking dining room filled with white linens, vivid flowers, and the glint of glass in the candlelight. My table was in the back, which allowed me a view of the entire room.

My fellow tablemates had come from around the world. There was a family of three from Brazil. Their daughter sat to the left of me, and we chatted during our meal. She also has FSHD and utilizes a wheelchair. She was a character; her personality shone bright. One thing we had in common was a love of piercings. We had a lot of fun. There was a lovely woman from France who sat across the table from me, and there were several researchers working on various projects for FSHD.

Dinner conversations turned into muted whispers as the FSH Society played a heartwarming short documentary about living without limits while diagnosed with FSHD. The young woman featured in this mini-film was vivacious and strong, a perfect embodiment of an FSHD fighter. She shared with us glimpses of her athletic lifestyle and how a positive attitude can motivate you into doing anything you set your mind on. It was lovely.

The auction followed, and two 2017 Super Bowl tickets were snatched up by a New England Patriots fan, raising \$8,500 for research toward a cure. The award ceremony came next (see story on page 16). Dan Perez was presented with a commendation signed by Governor Charlie Baker, who declared November 11 “Dan Perez and FSH Society



“ My fellow tablemates had come from around the world. There was a family of three from Brazil. Their daughter sat to the left of me, and we chatted during our meal. She also has FSHD and utilizes a wheelchair. She was a character; her personality shone bright. ”

—TRISHA SPRAYBERRY

Day” in the state of Massachusetts. Thank you, Dan, for your 25 years of commitment to our disease.

Pianist Steven Blier and singer Miles Mykkanen entertained us with a rousing, beautiful program. And then the evening was winding down. Gala guests began making their last laps around the room saying their goodbyes. I called for an accessible taxi, figuring I might have a long wait. To my

surprise, my ride arrived within 15 minutes, and I was back at my hotel room just before 11 p.m.

I arose Saturday morning eager to get to the second day of the patient conference. My flight home to Las Vegas wasn't until after 5 p.m. that evening, so I figured I could get a couple of hours in at the conference before I headed for the airport to catch my flight.

That was my plan, but my taxi ended up arriving almost three hours late. The concierge at my hotel tried calling every taxi company in the city, but there was not an accessible taxi to be found. When one finally arrived, the driver spent 25 minutes futzing with a balky ramp. At that point, I decided not to risk missing my flight and headed to the airport.

Now, I could bore you with the details of spending four hours at Logan Airport waiting for my flight, but I think you can picture on your own staring out the window watching planes landing and taking off, wheeling back and forth through the terminal, playing with my phone, and grabbing a bite to eat from a food stand next to my gate before being strapped back into that narrow metal-framed aisle seat like an infamous serial killer and pulled backwards onto the plane.

Five hours later, I was back in Las Vegas, back with my family, and already anxiously looking forward to the next patient conference.

Most importantly, I did it!!! 



Johnny and June Cash, Patti Smith, and everyone in between. Max Adler, Wendy Liebman, and some of our volunteer Lovelies welcomed the livestream audience on camera downstairs as jazz pianist Christopher Dawson entertained upstairs, while our other Lovelies interviewed guests at the silent auction.

Dean Mora's band then beckoned everyone to descend to the ballroom for dinner and a heartfelt welcome from FSH Society Executive Director June Kinoshita and a couple of pithy sets from Wendy Liebman, who then turned the mic over to the amazing Zack Krone, who artfully encouraged ever-higher bids for the Super

Bowl tickets, holidays in the Hamptons and Pebble Beach, and more.

Central to the evening, the Fund-a-Cure appeal came next. Scientist Amanda Rickard reported on the year's inspiring advancements in clinical trials and progress toward finding a cure for FSH muscular dystrophy. The host committee's Ellen Rennell delivered a deeply moving portrayal of the challenges of living with and providing care for her husband Jeff, who is affected by FSHD. The response was astounding: a standing ovation and \$20,000 raised within four minutes.

The evening was capped with a rollicking costume contest judged by celebrities

Max Adler, Stephen Kramer Glickman, Josh Sussman, and Josh Kelly. Guests strutted their stuff while Dean Mora's band turned up the beat for dancing with music from swing to rock 'n' roll.

All in all, A Ghostly Gala 2 was a huge success! The event grossed nearly \$90,000 to support the FSH Society. Thanks to the livestream, public awareness outside of the FSH community has been expanded tremendously.

Griff Entertainment has edited a fabulous sizzle reel on A Ghostly Gala, which you can enjoy here: <http://griffentertainment.com/projects/ghostly-gala-livestream/>. 



Charity Navigator awards the FSH Society with highest rating for ninth consecutive year

Top 1 percent of US charities

by JUNE KINOSHITA
FSH Society

The FSH Society has received its ninth consecutive Charity Navigator four-star rating. Only 1 percent of charities reviewed have received nine or more consecutive four-star evaluations, indicating that the FSH Society outperformed most other charities in America.

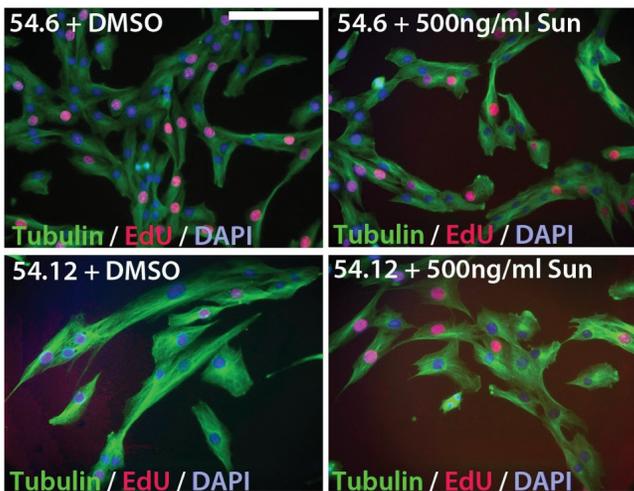


“It’s an honor to be recognized as a top charity in the country for the ninth consecutive year,” said Daniel Perez, president and CEO of the FSH Society. “This coveted four-star charity recognition is validation to donors that we are responsible with their financial donations and committed to our mission to generate greater awareness of the FSH Society and get one step closer to finding a cure for FSHD.”

The four-star rating from Charity Navigator reflects the FSH Society’s high standards for good governance and other best practices, as well as the ability to consistently execute its mission in a fiscally responsible way.

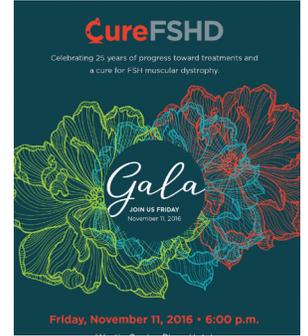
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CANCER DRUG AS A POSSIBLE FSHD THERAPY?



Sunitinib improves the pathogenic phenotype of FSHD myoblasts. (Figure courtesy of Zammit lab.) Myoblast (muscle precursor) cells from an FSHD “mosaic” patient (with a mix of normal and FSHD cells) were made into cell lines and labeled 54.12 (with FSHD genetics) and 54.6 (healthy control). Cells are labeled with tubulin protein (green) and DNA (red) to observe cell shape and cell division. Cell nuclei are blue. When sunitinib was added to healthy cells (top right), there was no effect compared to a control treatment with DMSO (top left). In contrast, 54.12 (FSHD) cells are long and thin when only DMSO is added (lower left), but become more similar to the 54.6 cells when sunitinib is added, suggesting that sunitinib is able to make the 54.12 cells appear more “normal.” The 54.12 cells also show more proliferation (purple nuclei) when sunitinib is added.

CureFSHD gala celebrates 25 years of the FSH Society



More than 250 people—individuals with FSHD, family members and friends, researchers, corporate donors, and other supporters—gathered at the Westin Copley Place Hotel in Boston on November 11, 2016, to celebrate 25 years of transformative achievements by the FSH Society. From top to bottom: Beth Johnston and Robyn O’Leary share a smile. Host Bob Oakes interviews Sarah Geissler as Steven Blier looks on. Silvère van der Maarel, holds his award (in the turquoise Tiffany’s box), chats with Michael Altherr and Alberto Rosa. Guests enjoy a chance to reconnect.