

FSH Watch



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



NETWORKING

2014 FSHD CONNECT

Hope to see you there!

We are delighted to announce that the FSH Society's biennial international patient, clinician, and researcher networking meeting will take place Saturday and Sunday, August 16 and 17, 2014, at the Westin Boston Waterfront Hotel, 425 Summer Street, Boston, Massachusetts.

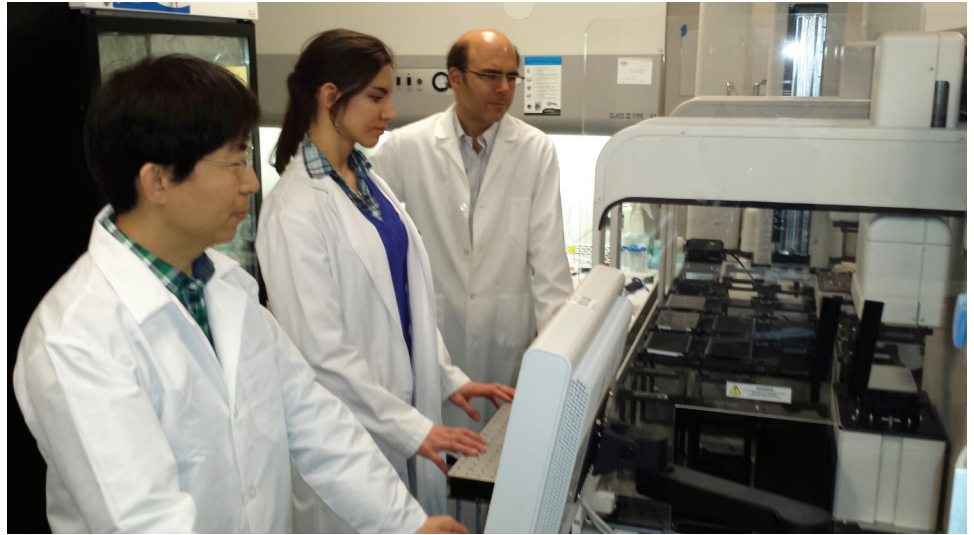
This year's meeting will bring together hundreds of patients and family members with top researchers, doctors, and health experts for two days of immersive learning and community building.

Attendees will have a wealth of opportunities to learn from some of the world's leading FSHD scientists about the cutting edge of research; have expert clinicians address specific health and medical questions; share valuable knowledge, insight, and support with fellow patients and families; and see old friends and make new ones. Popular breakout sessions include topics that members have requested, including

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Boston skyline overlooking the Charles River



Si Ho Choi, Natalie Schennum, and Michael Kyba (from left to right) keeping a close eye on the liquid-handling robot used for running high-throughput screens in their laboratory.



RESEARCH

First FSHD High-Throughput Drug Discovery Study

A Minnesota team has identified drugs that block the toxic effects of DUX4

Adapted from PRWeb

FSHD is among the most common muscle-wasting diseases, affecting more than 500,000 people around the world. There is no treatment today, but in a study published in the journal *Skeletal Muscle* on February 1, 2014, researchers have identified dozens of compounds showing early promise for future treatments.

"This is the first published high-throughput drug screening study for FSHD," noted June Kinoshita, Executive Director of the FSH Society, which helped fund the research. "Years of investment

in basic research to understand the genetic mechanism of the disease and to develop cell-based assays have made it possible to carry out this efficient strategy to identify drug candidates."

Recent discoveries point to a mysterious protein called DUX4 as a lead suspect in causing FSHD. Normally suppressed in adult muscles, DUX4 is unleashed in FSHD, with toxic effects on muscle cells. In people with FSHD, the facial (facio), shoulder (scapula), and upper arm (humeral) muscles are prone to degenerate, giving the disease its name. FSHD can also affect other muscles, including those of the lower abdomen and legs, leading to profound disability.

To hunt for drugs that can stop DUX4, the research team, led by Michael Kyba, PhD, of the University of Minnesota, engineered

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Why Does FSHD Target Muscles?



Signs of Spring

The flowering of FSHD research

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SPRING 2014

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June Kinoshita

Dear Friends,
 I recently traveled to our nation's capital to attend a meeting of the Muscular Dystrophy Coordinating Committee (MDCC). As my taxi sped past the Tidal Basin on the way to Bethesda, I saw that the cherry blossoms were on the verge of bursting into bloom, and I reflected on the numerous trips and more than 40 congressional testimonies that our Society's President & CEO Daniel Perez has made over the past 20 years to advocate for federal support for muscular dystrophy research.

This arduous work led to the passage of the landmark Muscular Dystrophy Community Assistance, Research and Education (MD-CARE) Act in 2001, which for the first time mandated that the US government fund research on all of the muscular dystrophies, including FSHD.

At the MDCC meeting, I witnessed just how profound an impact this legislation has had. Duchenne muscular dystrophy, with a half-century lead thanks to substantial non-governmental support from the Muscular Dystrophy Association, is furthest along with some dozen treatment trials ongoing.

But FSHD is not far behind! Fueled by your gifts to the FSH Society and your tax dollars channeled through the NIH, the genetic mechanism was discovered less than four years ago, and already the first high-through-

put drug screening study has been published (see our cover story in this issue). At our most recent international research conference, big pharma and biotechs were present in numbers not seen before.

I was also impressed by how the diverse muscular dystrophy communities can support one another. The pioneering efforts of Duchenne advocates are rich in lessons to instruct all of us. A rising tide truly lifts all boats.

The MD-CARE Act is up for reauthorization this summer, and I urge every one of you to write or call your senators and congressional representatives. Please let them know how well tax dollars have been spent to achieve progress in FSHD research, and urge them to sign on as sponsors of the bill. Our story on page 7 provides useful links.

You should take great pride in what your gifts and volunteering have accomplished. The Society's outreach programs and seed grants launched worldwide research efforts that are game changing. This very success now demands that we all do even more, to push forward, through ever increased funding and patient and family involvement in research, until we reach our goal of treatments and better lives for all patients.

With great hope and gratitude,

June Kinoshita
 FSH Society Executive Director

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

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The FSH Society Welcomes New Board of Directors Members

With diverse skills and a shared commitment to FSHD, the following new Board of Directors members bring welcome expertise:

AMY Z. BEKIER, MS

Amy obtained an MS in Education from Queens College, New York, and is retired from a successful career as an insurance agent and Chartered Life Underwriter. She utilized her teaching skills to train other agents. During this time she served on several boards and gained the knowledge and love of “giving back” to the community.

By including the FSH Society in her will, Amy is an FSH Society “Legacy Circle” member. As a Board member, she would like to help the Society expand its role in raising awareness of FSHD, which in turn will assist in patient identification and fundraising. Her personal motto, “I can no longer sit and cope but must get active and hope,” is her mantra and, hopefully, serves to inspire others.

Amy is an award-winning artist and avid golfer. When FSHD started to interfere with her passions, she found alternative ways of continuing to do the things she loves. Amy taught herself to paint left-handed when she could no longer raise her right arm. She enjoys painting with soft pastel and oil. Amy still plays modified recreational golf and is a member of the Bernardo Heights Country Club. She lives on the fairway of her home course in San Diego, California.



Amy Z. Bekier

ELLEN HANNAN, MBA

Ellen graduated from Skidmore College with a BS in Business and received an MBA from Pace University. She is a recently retired Wall Street analyst with over 30 years of experience in the energy industry. Ellen will co-chair the Society’s annual New York fundraiser, “A Festive Evening of Song.”

Ellen serves on the Board of Directors of Cinco Resources, Inc. as a member of the Audit, Compensation and Nominating committees. Her community service activities include serving on the Board of Trustees of Newton Country Day School of the Sacred Heart for over 15 years, where she headed the Investment Committee. She also served as a member of the Alumni Association Board for Skidmore College as the Chair for Career and Professional Development and is a past member of Skidmore’s



Ellen Hannan

President’s Advisory Council. Ellen continues to be active as Fund Chair and Class Agent for her class, and she is an avid bridge player.

As someone with a strong family presence of this disease, she has a deep, personal interest in advancing the efforts to raise awareness and fund the research necessary to find a cure for FSHD. She and her husband, Kevin Monahan, reside in Old Greenwich, Connecticut.

LEE F. (FRANK) KOLAKOWSKI, PHD

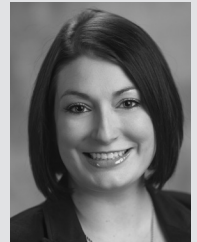
Frank is a Senior Scientist at Tetracore, Inc., a Rockville, Maryland-based biotechnology company that develops innovative diagnostic tools and instruments for the detection of infectious diseases and bioterrorism threat agents. Frank’s 25-year scientific research career also includes positions on the faculty of the University of Texas Health Science Center and as consultant for several US government agencies. He is currently focused on bioinformatics applications for genomics. Frank’s interest in scientific progress on FSHD stems from the disease’s impact on several family members. He holds a PhD in biological chemistry from the University of Pennsylvania and is the co-author of more than 40 peer-reviewed research articles on cell signaling and molecular biology. Frank enjoys photography, camping, and hiking. He and his wife Ann live in Timonium, Maryland.



Lee F. Kolakowski

LINDA LAURELLO-BAMBARGER

Linda is the Chief Financial Officer of Delta Railroad Construction, Inc. and was previously Project Manager and Contracts Manager for Delta. She is a graduate of John Carroll University with a BS in Business Administration and is currently finishing her MBA in Finance at Northeastern University. In January of 2012, she lost her grandfather, Cosmo (Larry) Laurello, who had FSHD and was a long-time board member and major supporter of the FSH Society. Her family history of FSHD, coupled with her love and admiration for her grandfather Larry, gives her a passion for helping to find a cure for this disease. She is excited to help fight for a cure and to be an advocate for patients and families. Linda and her husband, Shane Bambarger, live in Jefferson, Ohio. FSHWatch



Linda Laurello-Bambarger

2014 FSHD CONNECT

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teenagers and young adults, caregiving, traveling, and sports.

Please visit our website at www.fshsociety.org and download the registration form or register online no later than August 1, 2014.

Overnight accommodations are available at the Westin Boston Waterfront. The FSH Society has a special conference rate of \$189 per night (single or double occupancy), plus taxes. The hotel guarantees 30 accessible rooms, 14 with roll-in showers. For the best selection of accessible rooms and showers, make your reservations early. The closing date for reserving from the Society’s block of rooms is Tuesday, July 22. Please indicate your Group Affiliation as “FSH Society” when reserv-

ing. For reservations, call 888-627-7115 or 617-532-4600, or reserve online at: <https://www.starwoodmeeting.com/Book/FSHSociety>. FSHWatch

Registration

EARLY BIRD REGISTRATION BY FRIDAY, JULY 11: \$190 per adult (FSH Society member), \$245 non-member, \$130 per young adult age 12-18, and no charge for children under 12.

REGISTRATION BY FRIDAY, AUGUST 1: \$225 per adult (FSH Society member), \$275 non-member, \$150 per young adult age 12-18, and no charge for children under 12.

A Lifetime of FSHD

A patient and mother speaks at congressional briefing



On February 25, 2014, Lilleen Walters, an FSHD patient and mother of a young son with FSHD, spoke at a US Senate briefing on the reauthorization of the Muscular Dystrophy CARE Act. We thank Lilleen for making her remarks available to share with you.

Good afternoon honorable Senators, guests, and friends. My name is Lilleen Walters and I have facioscapulohumeral muscular dystrophy, or FSHD. I want to share with you an area where I'm an expert, painting a picture for you of what a lifetime of living with FSHD muscular dystrophy looks like.

It's much easier to say FSHD than the three muscle groups that this muscular dystrophy typically affects. Actually, it's a devastating and destructive disease that destroys every skeletal muscle in your body. The onset and severity vary widely among those affected with FSHD. A half-million people worldwide have this disease.

Appearance equals reality; our society is driven by impression. Consider the consequences of facial weakness—making friends, job interviews—they all become difficult if people are confused by your lack of facial expression. When you first meet people, you want to greet them with a big smile. I'd love to greet you with a big smile, but FSHD doesn't allow for that. No one should underestimate how awful this is.

At the age of seven, I didn't smile for a wedding picture no matter how hard I tried. My parents took me to doctors because I didn't smile. I was diagnosed and treated for depression, and my parents were assured everything would be fine.

At 15, I had a fall and injured my hip. I wasn't recovering; I was getting weaker, and no one understood why. After several hospitals and numerous tests I got the news: I had a rare type of muscular dystrophy—FSHD. What a dramatic change from my previous misdiagnosis of depression!

One doctor told me I had a slow, progressive disease with no impact, while another told me I would be in a wheelchair by the time I was 30. That's a lot for a teenager to comprehend, wouldn't you agree? FSHD isn't understood by most people in the medical community and is often misdiagnosed as it was with me.

Twelve years later, I was expecting my first child. While most mothers would be happy, I was concerned that my child would inherit FSHD from me. There was no genetic

“*At age five, Collin came to me and asked me why he couldn't smile. This was the first time I had to explain to my son that he had FSHD, and what that could mean for him and his future. It was incredibly painful to see him lose friends over the years because he couldn't physically go out and play like the other kids could.*”

LILLEEN WALTERS,
TESTIFYING AT US SENATE BRIEFING



testing or newborn screening available. Since I thought my disease only affects adults, I reasoned that if my child were to have it, at least he would have a normal childhood.

When Collin was born in November 1996, I was optimistic because he had a big smile. God had blessed us with a child free of the disease! However, within a year that smile disappeared, and we knew that Collin had FSHD. What we didn't know was how severe infantile-onset FSHD would be.

At age five, Collin came to me and asked me why he couldn't smile. This was the first time I had to explain to my son that he had FSHD, and what that could mean for him and his future. It was incredibly painful to see him lose friends over the years because he couldn't physically go out and play like the other kids could.

We looked for things that Collin could do. He joined Tae Kwon Do, and like everything he does, he attacked it and received his black belt. That was five years ago. Since then, I've watched helplessly as Collin went from being an outgoing, friendly and active child to an introverted teen who stays home focusing on school and playing video games.

Collin is now a junior in high school. Although we've had our challenges with the school system, he has a 3.7 GPA, taking all

honors and advanced placement classes. Collin continues to walk even though his doctors have told me that by all medical standards he shouldn't be able to do so due to his muscle weakness. Collin does it out of sheer determination.


The MD-CARE Act has been instrumental in providing funding for research and has resulted in many breakthroughs. My son Collin and I live with a disease that is slowly destroying our bodies, and we continue to hope that a treatment is found in our lifetime. But without continued funding through the MD-CARE Act, research will be eliminated and with it, the hope for a better tomorrow.

On behalf of the FSHD community, I strongly urge the continued support of the MD-CARE Act. The Congress and NIH have helped make unprecedented and remarkable progress with coordinated investments in research. Thank you for making these gains possible.

We must continue. If not us, then who? And if not now, then when? I believe that together, it's time we do something! With reauthorization and modest investments we can restore a lot of smiles to a lot of people. Thank you very much for allowing me to share our story with you. [FSH Watch](#)

“The Momentum Is Building”

MD-CARE Act Reauthorization Briefing



On February 25, 2014, Kathryn Wagner, MD, PhD, Director of the Center for Genetic Muscle Disorders at the Kennedy Krieger Institute in Baltimore, Maryland, and FSH Society Scientific Advisory Board member, spoke at a US Senate briefing on the reauthorization of the Muscular Dystrophy CARE Act. We are grateful to her for making her remarks available to share with you.

Honorable Senators, Staff, and the Muscular Dystrophy Community,

Mrs. Walters has given you a perfect description of facioscapulohumeral muscular dystrophy. It is a rare disorder, but one of the most common muscular dystrophies, affecting approximately one in 15,000 of the US population. It is a genetic disorder, frequently affecting multiple members of a family, inherited from one generation to the next. It is a chronic and disabling disorder, causing progressive and irreversible weakness of the face, arms, trunk, and legs. Pain and fatigue are frequent symptoms. It is currently an incurable and untreatable disorder. There are no accepted pharmacological treatments to slow the progression of the disease. Treatments are limited to supportive care such as orthopedic bracing, physiotherapy, and pain management.

Until recently, FSHD was also an enigmatic disease. It has been known since the 1990s that a contraction of repetitive elements on the tip of the long arm of chromosome 4 was linked to the disease. But how this contraction led to disease was unknown. The lack of an understanding of the pathophysiology of the disease, the lack of patient materials such as cells and muscle samples, and the lack of animal models stymied the field from moving forward.

This began to change in the early 2000s. Increased funding for FSHD led to an understanding that the chromosome 4 contraction causes misexpression of genes which are normally silent. The DUX4 retrogene becomes aberrantly expressed in skeletal muscle and turns on a battery of other genes. The MD-CARE Act ensured the funding of a Senator Paul D. Wellstone Muscular Dystrophy Cooperative Research Center in 2008 focused on FSHD.

In the first five-year cycle of funding, this Wellstone Center established a biorepository of DNA, cells, and muscle tissue samples from over 50 FSHD patients and their first-degree, unaffected family members. It was a tremendous outpouring from the FSHD community, each [person] literally donating a piece of him or herself to provide investigators with the necessary tools. These biomaterial tools are now available to all FSHD researchers throughout the world.

Using the muscle biopsy samples, genes expressed in FSHD but not in unaffected family members were identified and are considered “biomarkers” of the disease that can now be followed in future pre-clinical and clinical studies.

Animal models of FSHD have been recently developed and published over the last year, including a zebrafish model expressing DUX4, a transgenic mouse model expressing a portion of the abnormal human chromosome 4, and a xenograft mouse model transplanted with human FSHD muscle.

The Wellstone Center focus on FSHD was recently renewed. The Center, as well as other groups across the country, are now well poised to take advantage of these recent advancements in the under-



Kathryn Wagner

standing of the pathophysiology, the biorepository of FSHD tissues, and the new animal models to develop meaningful treatments for FSHD.

Multiple groups funded by the NIH are developing novel therapeutic initiatives for FSHD, with the goal of reducing the expression of the DUX4 gene or its downstream targets, and improving strength and quality of life. For the first time, a high-throughput drug screen in FSHD cells is underway. Gene therapy studies to knock down DUX4 expression in the new animal models have also begun.


These are exciting and new preclinical initiatives, to develop therapies directed at the specific pathophysiology of FSHD. The next step will be translating these preclinical discoveries to pharmacologic and gene therapy clinical trials in FSHD patients.

The field is preparing for this imminent reality by defining biomarkers of disease that can be followed in clinical trials including molecular biomarkers, imaging (such as MRI) biomarkers, and functional and quality-of-life outcome measures. These current studies are only possible due to the investment the NIH has made in FSHD. The future clinical studies are only possible with continued support such as the MD-CARE Act and through partnership with industry.

Industry is becoming interested in FSHD now for the first time. Previously, without an understanding of the pathophysiology and without animal models, there had only been one clinical trial with a novel drug and only a half-dozen trials total in FSHD. Now, with the recent advances, pharmaceutical companies have partnered with those in academics in providing drug libraries to screen and developing novel therapeutics targeted to the misexpressed genes.


The momentum is building to bring novel therapies into the clinic for FSHD patients. This next step will require expanding funding to capitalize on the advances made since initiation of the MD-CARE Act and expanding a currently limited workforce. There are simply not enough people working on muscular dystrophy due to limited funding. Through the Wellstone Centers, the MD-CARE Act has provided funding to support and educate trainees. We are training a new cadre of muscular dystrophy clinical and basic researchers to go further and provide meaningful therapies to FSHD families.

The patients that you have heard from today, the patients and family members in the audience, and the larger muscular dystrophy community at home and at work are looking to their federal government for a meaningful treatment for their disease. The investment has been initiated, but the payoff has not yet been realized. None of the muscular dystrophies you have heard about today have any meaningful treatment.

The families are not only dealing with the burden of their disease, but they are raising money for research, participating in clinical trials, and donating their tissue. Industry is partnering and spending a large amount of resources on developing novel therapeutics for these diseases. But there is no substitute for the MD-CARE Act, not only in NIH funding but in guidance. On behalf of the muscular dystrophy community, I ask that you reauthorize the MD-CARE Act. 

Speaking Up for FSHD Inside the Beltway

FSH Society members attend MDCC meeting

 The Muscular Dystrophy Coordinating Committee, a body mandated by the US government to review and coordinate federal initiatives for muscular dystrophy, met on August 26, 2013, at the National Institutes of Health in Rockville, Maryland. In attendance to represent the interests of FSHD patients was a group convened by the FSH Society that included the Society's Washington, DC counsel Morgan Downey and Society members Don Burke, Max Teleki, and the Walters family—Collin, Gary, and Lileen. Don Burke spoke about the specific needs of the FSHD community. Below is a lightly edited transcript of his unscripted remarks.

The first thing I want to do is thank all of you who are on the MDCC for all that you are doing. I have been pretty much involved in FSHD-related issues for more than 25 years, and the progress we have made since the 1980s is really pretty spectacular. So please keep it up.

Today's conversation has really felt like it was all about Duchenne, with once in a while a little bit about something else. There is a large contingent of us who live with adult forms of muscular dystrophy, and I'd really like to emphasize that. The challenges we face are very different from those of Duchenne. I couldn't help but think about my life and the lives of people around me as Annie Kennedy [of the Muscular Dystrophy Association] was giving her talk about transitions into adulthood.

For those of us with adult forms of muscular dystrophy, we face the opposite challenge. So we reach adulthood, and then we are faced with this unknown: When are we going to lose the ability to care for our families? When are we going to lose the ability to ambulate or to do our jobs?

I can't tell you how many people I've talked to who were in jobs that required a great deal of physical exertion, and then one day their disease just took over and they couldn't do their jobs anymore. There are



Don Burke

dystrophies. I think that's particularly true with FSHD, as we have sort of narrowed in on DUX4 as a candidate for the disease.

I think, from all of the reading I'm doing, it's important to maybe step back and ask, Do we really understand what is in FSHD and what is not in FSHD? Do we understand the genotype? Do we understand the haplotype? Why are there cases of the haplotype, but no phenotype for the disease? These are really important questions to ask, and I worry that we are getting too narrowly

sequencing, and so forth. We heard multiple speakers talk about prevalence today, of not understanding the true prevalence of these diseases in our society because so many people never report them or are misdiagnosed. Are there novel ways that we can get at prevalence? Can we use the modern, connected world that we're in today to better understand prevalence, ideally even at the genome level?

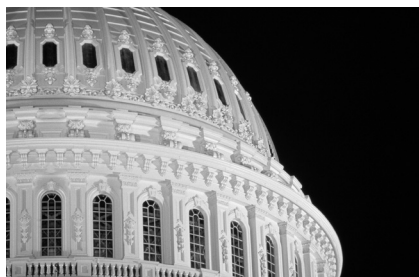
The military talked a little bit earlier today about what they were doing with Duchenne muscular dystrophy, but they are also very active in the Wounded Warrior Program, coming up with really amazing technological solutions to devastating injuries. Are there ways that we could leverage some of that work, which ranges from muscle regeneration to advanced powered exoskeletons?

I would encourage this group to consider these as you look at your action plan going forward, because until we have cures, all of us need to live with these diseases. As we go forward, technology and novel approaches to compensating for the disease are really important. In FSHD, as we lose our bodies, there is virtually nothing, short of scooters, that we have available to us as compensating devices.

Thank you very much for the opportunity to speak to you today. [FSHWatch](#)

“ I'd also like to encourage this group to think about novel ways of approaching the research related to FSHD and other muscular dystrophies. ”

DON BURKE,
TESTIFYING AT US SENATE BRIEFING



very real consequences to the adult side of the disease as well. And FSHD in particular is not just an adult disease, as is obvious here today.

John Porter [from the National Institute of Neurological Disease and Stroke] mentioned the need to step back a little bit and reassess where we are with some of these muscular

focused on DUX4 as sort of the miracle solution to the disease.

I'd also like to encourage this group to think about novel ways of approaching the research related to FSHD and other muscular dystrophies. There is a lot going on with genomics, the ability to do whole-genome

FIRST FSHD HIGH-THROUGHPUT DRUG DISCOVERY STUDY

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mouse myoblasts (immature muscle cells) to express DUX4 under the control of a genetic switch that is triggered by adding the antibiotic doxycycline to the petri dish.

When DUX4 is switched on, the cells begin to die, and they also become more vulnerable to a variety of chemical insults. Drugs were added to the cultured cells to see if any of them rescued the cells.

The research team tested thousands of compounds on these DUX4-expressing cells, including 1,120 Food and Drug Administration-approved off-patent drugs and 43,000 other chemicals. After extensive studies to weed out false positives, and further winnowing to select compounds with favorable chemical properties, the investigators honed the list


to 52 “hits,” or candidates.

“Remarkably, two-thirds of our hits are compounds that protect cells from oxidative stress,” Kyba said. “Although we need to be cautious extrapolating from cells in a dish to human patients, I am enthusiastic about testing whether protecting cells from oxidative stress is beneficial in FSHD.”

Further work is now being planned to understand the precise mechanism behind the anti-DUX4 activity of each of these compounds. This research will help the investigators focus on the most promising ones to develop into therapies. At the same time, each compound can bring to light new insights into how DUX4 causes this devastating disease.

“We were very, very fortunate to get

support from the FSH Society in the form of a postdoctoral fellowship back in the dark ages when nobody else was funding research on DUX4,” said Kyba. Additional funding came from the National Institutes of Health; Dr. Bob and Jean Smith Foundation; Friends of FSH Research; the FSHD Global Research Foundation; and the Muscular Dystrophy Association.

For more information about Dr. Kyba’s work at the Lillehei Heart Institute at the University of Minnesota, visit <http://www.tc.umn.edu/~lhi/kybalab/> 

Reference

Bosnakovski D, Choi SH, Strasser JM, Toso EA, Walters MA, Kyba M. High-throughput screening identifies inhibitors of DUX4-induced myoblast toxicity. *Skeletal Muscle*. 2014 Feb 1;4(1):4.

MD-CARE Act

WRITE TO YOUR SENATORS AND CONGRESSIONAL REPRESENTATIVES

The FSH Society is calling on its members and supporters to call or write to their senators and congressional representative and urge their support for S. 315 and H.R. 594—bills that would reauthorize the Paul D. Wellstone Muscular Dystrophy Community Assistance, Research and Education Act (known as the MD-CARE Act).

Research on FSHD has received a considerable boost since the enactment of the MD-CARE Act in 2001. The Act was reauthorized in 2008 and is now up for renewal again. If not reauthorized by Congress, parts of the Act will expire.

The Act provides for the establishment of Paul D. Wellstone Centers of Excellence, several of which are focused on FSHD, epidemiological tracking by the Centers for Disease Control and Prevention, and the establishment of the Muscular Dystrophy Coordinating Committee, which brings together representatives from several government agencies as well as from patient groups to develop comprehensive strategies on research and related topics. (For many years Daniel Perez of the FSH Society served on this committee.)

It is important to have as many members of the House and Senate as possible co-sponsor the legislation in their respective chambers. You can check two websites below to see which members of Congress are already co-sponsors. If they are co-sponsors, please call or write to them anyway to express your awareness and gratitude for their support.

Links:

The Thomas site (part of the Library of Congress) lets you follow the bills’ progress and see an updated list of co-sponsors. Enter “muscular dystrophy” to search by word or the bill numbers, H.R. 594 in the House of Representatives or S. 315 in the U.S. Senate.

<http://thomas.loc.gov/home/thomas.php>



The Muscular Dystrophy Association site contains a list of co-sponsors of the legislation in the House of Representatives and the Senate, as well as a timeline of accomplishments under the Act.

http://mda.org/advocacy/md_care_act

The Parent Project Muscular Dystrophy site has more details on the MD-CARE Act itself.

http://www.parentprojectmd.org/site/PageServer?pagename=Advocate_mdcare

Please note:

The MD-CARE Act does not set the amount of spending on FSHD or the other muscular dystrophies at the National Institutes of Health. Funding levels are set in the “appropriations process,” which technically is a separate legislative process. However, the reauthorization of the MD-CARE Act raises the visibility of all the muscular dystrophies, which is essential in the appropriations process. The Appropriations Committee and process establishes the amount of money the NIH receives each year from Congress to fund biomedical research.

Research Priorities for FSHD 2014

FSH Society conference sets agenda

The FSH Society has been convening an annual International Research Consortium and Research Planning conference since 1994. At the conclusion of each meeting, the attendees discuss the priorities for the next year. What follows is the summary from the most recent conference, held in Cambridge, Massachusetts, on October 21-22, 2013. We thank FSH Society Scientific Advisory Board members Michael Altherr, PhD; Rune Frants, PhD; David Housman, PhD (SAB chair); and Silvere van der Maarel, PhD; and FSH Society President Daniel Perez for compiling this summary.

DUX4. The unanimous conclusion of the general discussion was that overexpression of the toxic transcription factor DUX4 is at the root of FSHD1 and FSHD2. Expression of DUX4-fl mRNA (messenger RNA) and protein is dependent on two conditions: 1) a specific DUX4 haplotype containing a poly-A site and 2) an open chromatin structure, due to D4Z4 repeat contraction-dependent (FSHD1) or contraction-independent (FSHD2) mechanisms, the latter due to mutations in the SMCHD1 gene encoding a chromatin modulating enzyme. There are indications for further genetic heterogeneity, thus additional gene defects causing FSHD. The chromatin relaxation of the DUX4 region (close to the #4q telomere) can induce additional gene expression effects in cis (#4) and trans (other chromosomes). As DUX4 is a transcription factor, the overexpression can trigger a cascade of downstream molecular pathways contributing to the large variability in the clinical phenotype and natural history of FSHD. Conclusion: DUX4 expression is necessary but not always sufficient to cause FSHD. Research should focus on upstream and downstream molecular pathways and mechanisms, as they form the most plausible intervention targets.

Disease models. The field needs improved and specific in vivo (animal) models for mechanistic and intervention studies. At this stage it is not sensible to give strict recommendations. Inducible (conditional) models seem necessary to dissect spatial



Rune Frants poses a question at the FSH Society's International Research Consortium meeting.

The FSHD field is working hard to establish patient databases with detailed clinical and genetic information. Equally, the development of sensitive quantitative clinical monitoring methods to follow intervention trials has a high priority. It is important to closely follow the situation in related fields.

and temporal effects of the DUX4 pathway. For specific questions, simpler models, like zebrafish, may have unique potential. Various xenograft models aiming at generating human muscle in mouse muscle are promising, but strongly dependent on availability of human muscle biopsies or cell lines. In other muscle disease fields, AAV mediated “gene therapy” has proven its value. Availability of higher vertebrate models (e.g., dogs, primates, etc.) may be helpful to study intervention effects prior to human trials.

Intervention. Is there a magic bullet for FSHD treatment? Although DUX4 overexpression is crucial, various biological and chemical (pharma) strategies can be envisaged to intervene with the overall expression mechanism, directly by targeting the mRNA (messenger RNA) or the protein, or indirectly by modulating the transcription machinery, including the chromatin struc-

ture. As the ultimate goal is to influence the clinical phenotype and disease progression, intervention in specific molecular sub-pathways of the DUX4 cascade may form alternative strategies.

Clinical studies and trial readiness. There is an urgent need for better understanding of the natural history of FSHD, e.g., the clinical heterogeneity among and within families, the asymmetric expression of the disease, the cause and consequence of inflammation, and the effect of physical and cognitive training, etc. The FSHD field is working hard to establish patient databases with detailed clinical and genetic information. Equally, the development of sensitive quantitative clinical monitoring methods to follow intervention trials has a high priority. It is important to closely follow the situation in related fields, e.g., Duchenne muscular dystrophy.

Priorities for 2014

- The DUX4 interactome
- Understanding DUX4 manifestation and variation
- Additional genetic heterogeneity; non-FSHD1 and FSHD2
- Understanding pathophysiology of FSHD: connection to DUX4, heterogeneity, asymmetry, role of inflammation; infiltrates and etiology
- Disease models
- Well-documented natural history with reliable endpoints; modulating mechanisms/genes
- Increasing data depth of patient databases with extensive (follow-up) clinical data
- Preparation for clinical trials: reliable and meaningful outcome measures; access to discrete patient populations and disease mechanism-of-action classes
- Therapy; proof-of-principle experiments
- Focus on translational research; from clinic to bench and back FSHWatch

Editor's note: For a glossary of science terms, see page 30 of the *Summer/Fall 2013 FSH Watch Research Issue*, available at our website at <http://fshsociety.org/pages/comNewsletters.html>.

FSHD Research Gains Visibility

Report from the 2013 World Muscle Society Congress

by **LINDSAY WALLACE, PHD**
Columbus, Ohio

Last October, I had the privilege of attending the 18th International Congress of the World Muscle Society (WMS). The meeting was held at the Asilomar Conference Grounds in Pacific Grove, California.

The rustic atmosphere of the campgrounds and the peaceful dune walks along the beach promoted relaxation. However, there was a general unrest amongst conference attendees as the federal government shutdown began on the first day of the meeting. With this news came a barrage of program changes as some invited speakers, poster presenters, and others were required to leave the meeting and return home. Nevertheless, the rest of us powered on and had quite a successful meeting.

The opening ceremony as well as the general tone of the Congress is traditionally centered on the “Triple-E’s”: Education, Enjoyment, and Excitement. To kick things off, we learned about the history of the area from three local experts: a resident ranger, a professor from Cabrillo College, and a Native American Ohlone tribe descendant. In keeping with the theme of the meeting, they spoke on what the locals considered “the strongest muscle of them all”: the abalone. Following the opening ceremony, the shellfish settled for being the star of the dinner (which for me also fulfilled the last two “E’s”!), and talks were focused on neuromuscular topics.

The three selected categories of focus for the 2013 meeting were: 1) The Myomatrix in Health and Disease; 2) Immunity and Muscle Disease; and 3) Advances in Therapy for Neuromuscular Disorders.

There was a lot of buzz centered around the third category because just prior to the Congress, Prosensa announced that its Phase 3 clinical study for treatment of Duchenne muscular dystrophy with drisapersen had failed to meet the primary endpoint of the trial. The path forward for antisense oligonucleotides as well as discussions of the Six-Minute Walk Test for outcome measures were hot topics of the meeting. There were also some very promising results shared about therapies for spinal muscular atrophy from multiple groups, and industry symposia hosted by Genzyme and Sarepta Therapeutics. The mornings were filled with lectures and oral presentations, while the afternoons hosted interactive poster sessions. I am happy to report that following a very poor showing of FSHD research at the 2012 Congress, the 2013 Congress had three times as many FSHD presentations. Silvere van der Maarel reached the broadest audience with his invited lecture, “New Advances in Facioscapulohumeral Dystrophy.” This lecture was an overview of the genetics of FSHD



The Asilomar Conference Center hosted the World Muscle Society meeting in October 2013.

and a summary of the most recent discoveries in the field.

The fact that FSHD received one of the invited platform talks, in my opinion, shows the momentum of the research and a positive increase in visibility. Furthermore, van der Maarel very eloquently made the complexity of FSHD understandable for the entire audience, which is not an easy feat.


Van der Maarel’s talk laid the background for the rest of the FSHD presenters, who were featured in a poster category along with oculopharyngeal muscular dystrophy. Eleven presenters, including myself, shared our research covering a broad spectrum

of FSHD topics. There were presentations on basic biological/mechanistic aspects of DUX4’s role in FSHD; bio-behavioral studies including psychoanalysis impact, anxiety, and altered sleep in patients; gene expression and epigenetic studies; and magnetic resonance imaging and physical therapy evaluations. The multidisciplinary views embodied at the WMS provide valuable insight.

Like most people in my position, I focus day in and day out on my particular niche of research. However, during my poster presentation I spoke with clinicians, physical therapists, pharmaceutical representatives, professors, and other trainees. The exposure to the way other disciplines approach important research questions or therapeutic strategies is, in my view, one of the biggest benefits to this meeting.

The size of this Congress is also quite manageable, giving plenty of time to catch up with familiar faces. Meal times provided for some of the best FSHD research discussions of the week, thanks to our colleagues Peter and Takako Jones from the University of Massachusetts.

The FSHD representation at the WMS was greatly increased in 2013, thanks in large part to the efforts of the FSH Society. I believe we can do even better. I mentioned before that the motto of the WMS is the Triple-E’s—Education, Enjoyment, and Excitement. Let’s take that to heart and do our part to *Educate* the world muscle community about the incredible FSHD research going on. Let’s get *Excited* about the progress we are making, and *Enjoy* what’s to come in 2014.

The 19th International Congress of the World Muscle Society will be held in Berlin, Germany. Details can be found at www.wms2014.com. Our field grew from four posters in 2012 to 12 presentations in 2013. How many can we get this year? 

Editor’s note: Lindsay Wallace is a postdoctoral researcher at Nationwide Children’s Hospital in Columbus, Ohio.



Living With FSHD in Kenya

Struggle and hope

by **CHRIS CHEGE**
Thika, Kenya

I was born 47 years ago in a remote area in central Kenya. I grew up just like any other kid, doing all types of odd jobs at that time. My parents were farmers, and we used to pick coffee from our small farm, as it was the only cash crop in our village.

I started schooling at the age of seven and all was well, but as time passed I started noticing that I could not run as fast as my fellow pupils. Some used to joke about me, and some would even fight me and run because I was weak.

My class teacher used to beat me, accusing me of being lazy. My only hope was my parents, but they also accused me of being lazy. Physical education in my primary school was an ordeal because of my weakness. But when I completed primary school and joined secondary school, things moved from bad to worse.

I used to trek seven kilometers daily, and to me it seemed like a way of discouraging me from going to school. My parents didn't bother to take me to a hospital for a checkup, and everybody believed I was lazy.

After secondary school, my parents didn't bother to further my education. I thank God that I didn't turn to drugs, although I was stressed. I had lived for two years at home when my elder sister



Chris Chege with his mum.



Chris' wife Keziah graduating with her Bachelor's of Business Administration.

formers, capacitors, and circuit breakers.

After college, I joined the Kenya Power Company in 1993 as a craftsman. My condition was getting worse at this time, and my employer was always harsh with me. I had numerous transfers within the company, as everyone thought I was lazy.

In 1997 I married my lovely wife Keziah. She loves and supports me in everything. She means everything to me. Whenever I am traveling she must come with me.

After marriage, my wife advised me to seek medical attention from the leading hospital in Kenya. I was able to see a neurosurgeon who diagnosed that I was suffering from muscular dystrophy (MD). With the help of my wife, I was able to accept my condition. Due to this, I started seeking knowledge about MD, and that's how I landed on the FSH Society website.

God has blessed us with two lovely boys, Dan and Jude. I always pray for them because I know they might have the

same genes. How I wish God will save them from this misery!

Despite my condition, I try to make them happy and also to educate them about FSHD. They are very much informed about the changes that occur as the years come and go. I was determined to let them be informed so that they do not grow up ignorant about the condition as I was.



“ My class teacher used to beat me, accusing me of being lazy. My only hope was my parents, but they also accused me of being lazy. Physical education in my primary school was an ordeal because of my weakness. But when I completed primary school and joined secondary school, things moved from bad to worse. ”

CHRIS CHEGE

Beatrice applied for college in 1989. It was like a miracle to me because she applied to the college all by herself. Almost everybody was against her and thought it was a waste of time.

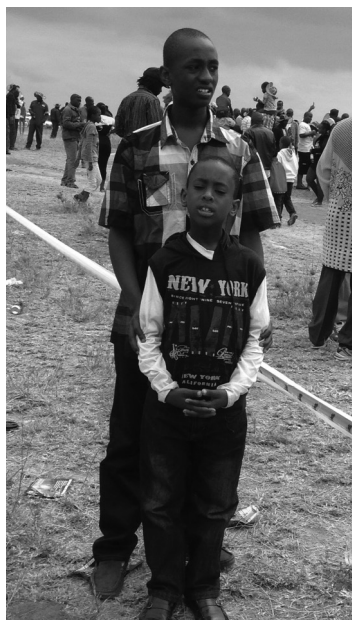
Beatrice helped me to apply to the electrical power training college. There, life was hard, with limited resources, but I kept on going. I was trained to construct power lines and install trans-



Soaring High

Rising to a new challenge

by **RAYMOND HUML**
Durham, North Carolina



Chris' sons Dan and Jude.



Chris with his sister Beatrice.

Here in Kenya, FSHD is not known to many. Most hospitals are not aware of this condition, and they don't have physiotherapy equipment.

For people with FSHD here in Kenya, life is harsh. Accessing most buildings is a nightmare. Government buildings—hospitals, courts, even parliament where laws are made—are not friendly to the disabled. So who is going to care, if the government does not?

Hotels also are not friendly, so I don't eat when traveling because even toilets are not accessible to the disabled. Transport is another nightmare because no single public service vehicle is friendly to my condition. When traveling by bus I have to hire two men to assist, and my wife must accompany me because I have frequent falls. Most people mistake me for a drunkard. It is very expensive for me to travel.

My mother, who suffers from the same condition, believes that I was bewitched together with my younger sister and brother, who are also affected. At one time my mum took us to our grandma's place for blessings, hoping the condition will heal. I have tried to educate her on FSHD, but the more I try, the more she believes I was bewitched.

Assisting others with the same condition is hard because of the lack of transport and funds. Also, most parents typically hide their children who have muscular dystrophy, thus making it even harder to notice them.

Here in Kenya, the National Council for Persons with Disabilities is the government body that deals with persons with disability, but it focuses mostly on the blind, the deaf, and those who lack some body parts. FSHD is simply not recognized.

My appeal to the FSH Society is to empower people from different parts of the world to educate others about this condition. For sure, I am ready for any clinical trial if it will improve the life of those suffering from muscular dystrophy.

God bless you all as we wait for a cure. **FSHWatch**

My son Jonathan R. Huml never was one to take the easy path, and if not challenged by others, he would challenge himself.

By the age of 12, he completed the requirements for his Eagle Scout rank. This achievement was the result of hundreds of hours of work on merit badges and community service projects.

He played in two baseball championships, one with South Durham Little League and one with his eighth grade class at Immaculata Catholic School, with the ICS Eagles. Playing catcher on the team with the best record at ICS in 10 years, he earned the only sports award given to students each spring: The Dedication and Determination Award.

Jon was diagnosed with FSHD at the University of North Carolina in 2013, 10 years after his sister was diagnosed with the same affliction. He didn't choose the FSHD challenge but, whether as an Eagle Scout or an Eagle on the ICS baseball team, Jon is determined to soar above it.

On December 30, 2013, with the encouragement and support of his grandfather Raymond G. Wile, Jon took his first Discovery flight in Williamsburg, Virginia, and was hooked. He now takes regular flying classes at Raleigh/Durham International Airport. He is surrounded by a team of professionals who are committed to helping Jon obtain his pilot's license.

When Jon's troop, 424, celebrated his Eagle Scout achievement in 2011 with a special Court of Honor, he was recognized by the *Herald Sun*, his local paper. The article contained a quote about Jon that rings true today: "When he sets his mind to getting something done, he does it." **FSHWatch**



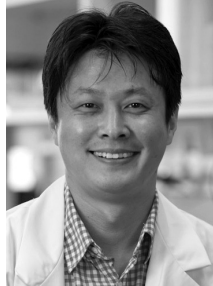
Jon Huml is getting ready to take to the air in this Cessna.

Stem Cell Research Update

Gabsang Lee at the Mid-Atlantic FSHD patient meeting

by **JIM FOX**
Bellingham, Washington

I was able to attend the Mid-Atlantic FSHD Support Group meeting at the Kennedy Krieger Institute in Baltimore on February 1, 2014. It was another good meeting, with some 25 FSHD folks, caregivers, researchers, and advocates attending, and dozens more watching a live video feed online.



Gabsang Lee, PhD

The speaker was Gabsang Lee, PhD, of Johns Hopkins Hospital's Department of Neurology, Institute for Cellular Engineering. His current work is partially funded by a grant from our FSH Society.

Lee spoke about his team's current research and development of a new way of producing skeletal muscle cells with stem cells for research (and eventual treatment). This breakthrough protocol promises significantly shorter development time and lower costs, with less ethical controversy, and improved quality and applicability in comparison to previous embryonic stem cell approaches.

Stem cells, with their ability to differentiate into various cell types (muscle, blood, nerves, skin, etc.), hold the promise of regenerating damaged tissues in a wide variety of injuries and diseases.

Embryos, by their very nature, are an obvious source for stem cells, but they present ethical and practical disadvantages. In addition, stem cells with unrelated DNA raise the risk of being rejected by the host patient's immune system. Recent research has focused on ways to obtain stem cells from patients themselves, based on the discovery that mature adult cells can be "reprogrammed" to rewind the genetic clock and become stem cells again. These are known as induced pluripotent stem cells (iPSCs).

Induced pluripotent stem cells can be differentiated into many cell types. Lee spoke about progress in his lab to produce iPSCs and, subsequently, skeletal muscle cells, in sufficient quality and quantity for research and eventual therapies. With the

ability to generate billions of cells, it should become possible to use the cells as a model system to screen for drugs.

To treat genetic disorders through stem cell transplants, the iPSCs would need to have the genetic problem fixed in the test tube before being translated into patients. Efforts are underway to "cure" FSHD in the test tube through various genomic engineering methods.

With a good supply of genetic material and cell lines now at the UMass Wellstone Center (donated by nearly 50 FSHD families), researchers are working to apply these resources to our specific FSHD issues.

As for timing, biomedical research is a long, complex process. If not on the cusp of a breakthrough, we are at least well into the beginning, with clear paths ahead. We must build the tools for research, and even after finding solutions, there will be years of safety testing before use on humans can be approved.

Thanks to the FSH Society's advocacy and participation by our members, we are seeing research and progress in these areas, producing exciting results, and holding promise for the future.

What we now need are increased funding and political advocacy, something each of us—patients, family, friends—can contribute as local groups and individuals. Let your vari-

ous representatives know that you want resources prioritized for this kind of beneficial research. Do what you can to raise finances and public awareness about FSHD and this kind of leading-edge research. Research findings for FSHD benefit everyone!

We also need volunteers for research or trials. Search ClinicalTrials.gov for studies that are seeking volunteers. Sign up with the National FSH Registry at the University of Rochester. We still need muscle tissue samples, especially from non-Caucasian families and families with members differing in onset and severity.

The video of Lee's talk is posted on the FSH Society's YouTube channel. After the presentation, we had some good informal discussions and social chats with old and new friends.

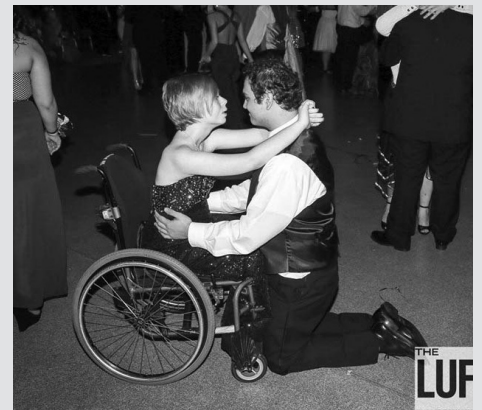
It was a good meeting. Thanks to all who put it together and who are working to solve the FSHD problem. I look forward to hearing about more areas of progress in upcoming Mid-Atlantic patient meetings this May 10 and September 6. [FSHWatch](#)

Editor's note: If you are interested in volunteering for research, please email Daniel Perez at daniel.perez@fshsociety.org. To learn about the National FSH Registry, go to <http://www.urmc.rochester.edu/neurology/national-registry/>.

Inspired by Shelby

Shorty Jackson and his friends in Lufkin, Texas, held a fundraiser last year. They raised over \$4,000 for FSHD research, inspired by Shorty's daughter Shelby (right), who was diagnosed with FSHD at age eight and is now 16. "As a parent, when you get news like that you quickly find yourself wanting to be involved as much as you can in finding a cure, raising awareness, or helping those affected," says Shorty. Thank you so much to the Jackson family and their friends who volunteered!

Photo of Shelby at her high school prom by Andy Adams, courtesy of *The Lufkin Daily News*.



Respiratory Involvement in FSHD

New study flags which patients are at greater risk

by JUNE KINOSHITA
FSH Society

Reduced lung capacity has a significant negative impact on a subset of FSHD patients, but there have been few data about which patients are most at risk. Now, Michelle Scully and her colleagues at the University of Rochester Medical Center have published the largest study to date, shedding important light on the subject.

The Rochester study recruited 61 volunteers with genetically confirmed FSHD, 53 with FSHD1, and eight with FSHD2. (FSHD1 is caused by deletion of D4Z4 units on chromosome 4; FSHD2 is caused by mutations in the gene SMCHD1.) More than 95 percent of FSHD patients have FSHD1. The range and severity of symptoms are the same for both types 1 and 2. The volunteers' mean age was 49.5 years, with onset typically in the third decade, and overall they reported being moderately affected.

Reduced lung capacity was observed in 9.8 percent of FSHD1 patients and in 25 percent of FSHD2 patients. "The higher frequency of restrictive lung disease in FSHD2 seen here requires confirmation in a larger cohort of FSHD2 patients," the study noted.

For most individuals with FSHD, however, the study's news is actually good. "I think we offer reassurance," said the study's senior investigator Jeffrey Statland, MD. "The majority of patients had no evidence for respiratory involvement. Indeed, the mean forced vital capacity for the group was around 100 percent of what is predicted for age and gender."

The patients who had reduced lung capacity were those who were also more severely affected by their disease. According to Statland, "The patients who we need to be concerned about lung involvement tended to use wheelchairs and have weakness in the muscles of their upper thighs or around their hips. We saw this in both FSHD Type 1 and Type 2."

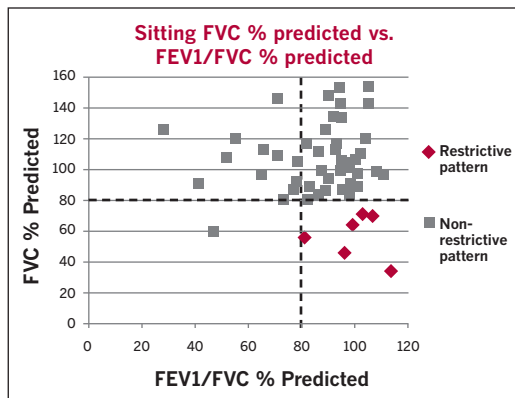
If an individual falls into this higher risk category, what then? Statland advised: "It is reasonable to screen these patients for respiratory involvement, at the minimum obtaining baseline pulmonary function measurement and considering checking bedside spirometry yearly.

"We generally screen patients who have prominent weakness of the trunk muscles or patients with prominent pectus excavatum [hollowed chest]," Statland added, although he said this study did not investigate these specific factors in relation to lung capacity. "Once someone is having to use a wheelchair at any point during the day," he added, "it is probably important to screen ... yearly" for respiratory function.

This study was supported by grants from the National Institutes of Health and the Muscular Dystrophy Association. The authors also thanked the patients and their family members who were the impetus for this study. [FSHWatch](#)

Reference

Scully MA, Eichinger KJ, Donlin-Smith CM, Tawil R, Statland JM. Restrictive Lung Involvement in Facioscapulothoracic Muscular Dystrophy. *Muscle Nerve*. 2014 Feb 23. doi: 10.1002/mus.24218.

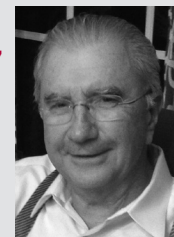


This graph depicts the lung capacity on the y-axis, and the FEV1/FVC ratio on the x-axis, which is a way to look for signs of obstruction, or difficulty exhaling all the air in the lungs. Most FSHD patients are in the right half (no obstruction). A subset of patients are in the bottom right quadrant, indicating that they have a reduction in their lung capacity. The majority are in the top right quadrant, indicating that their lung function is essentially normal.

Thank You, Bill Michael!

A WELL-DESERVED RETIREMENT FOR
FSH SOCIETY TREASURER

William G. (Bill) Michael, a founding member of the FSH Society, Board member and Treasurer since its inception in 1991, retired from these roles in February. A CPA since 1964, Bill was a partner in the public accounting firm Haskins & Sells (a predecessor to Deloitte) and, later, Managing Partner of Russell, Brier & Co. LLP, a Boston accounting firm, specializing first in audits of public corporations and thereafter in individual and estate taxation.



Bill Michael

Bill and his wife Ginny were the primary caregivers for their late son, William T. (Billy), who was greatly affected by FSHD and died in 2004 at age 35. They were devoted to Billy, ensuring that he continued to live with them instead of in an institution and helping enable him to live life fully.

As Treasurer and a director, Bill devoted countless hours to the Society, preparing and maintaining the internal accounts and financial statements, working with the independent auditors on the annual audit, reviewing tax and regulatory filings, advising on regulatory compliance, and educating and informing the Board about the Society's finances. Bill deserves much of the credit for the Society's sound financial footing and transparent operations. Since his official retirement, Bill has been mentoring the new Treasurer, Chris Stenmon, and creating a smooth transition.

Bill has been a personal friend and professional mentor to many of the members and directors of the Society, and especially to President & CEO Dan Perez. He has always been willing to give advice and support to people affected by FSHD and their families. Bill has testified eloquently and movingly at the NIH about the burden of FSHD.

The Society has established a research grant in Bill's honor.

A rated chess player, Bill plans to spend more time playing competitive chess. He and Ginny have a daughter, Beth, and three grandchildren.

—by Howard Chabner

Human Muscle Grows in Mice

A new research tool for FSH muscular dystrophy

by JUNE KINOSHITA
FSH Society

The FSH Society's outreach and support for research volunteers has enabled people with FSHD to donate muscle tissue, which scientists have succeeded in grafting into mice, providing a new tool for conquering this devastating muscle-wasting disease.

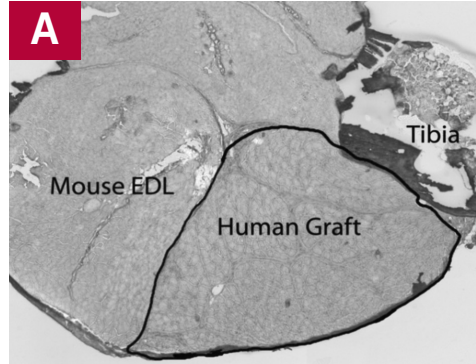
Since the discovery of FSHD's genetic mechanism in 2010, scientists have been forging ahead to find drugs and genetic therapies that could block this mechanism. But there remain major obstacles in the path to a treatment. One of the most significant roadblocks is the lack of a preclinical research model that can be used to study the disease in depth and to evaluate new therapies.

Building an FSHD model has proven to be a substantial challenge. The genetic mechanism of FSHD is extraordinarily complex, with components that do not exist in mice. To overcome this difficulty, a multi-institutional team led by Kathryn Wagner, MD, PhD, Director of the Center for Genetic Muscle Disorders at the Kennedy Krieger Institute and professor at The Johns Hopkins School of Medicine, decided to transplant human muscle into mice, grafting tissue taken surgically from the biceps of FSHD patients into the leg muscles in living mice.

The grafted muscles received a blood supply and nerve signals from the host mice, which were bred with defective immune systems to prevent rejection of the foreign tissue. The grafts survived for more than 40 weeks, during which time they regenerated. The grafted muscles could contract like normal muscle, and retained the cellular and genetic characteristics of muscle from a human with FSHD.

"Most potential novel therapies fail to successfully translate from animals to humans," says Wagner. "Growing human tissues in animals [xenografts] has previously led to the successful development of therapies for multiple cancers and now, with this new muscle xenograft model, we are hopeful that new therapies for muscular dystrophy will emerge."

The study's authors thanked the FSH Society for its "invaluable" help in recruiting FSHD patients to participate in the research. The Society helps to defray travel and lodg-



Cross-section from the leg of a xenograft mouse showing healthy human muscle growing alongside mouse muscle.

ing costs of individuals participating in the research. Study co-authors came from the Kennedy Krieger Institute, Baltimore, Maryland; University of Massachusetts Medical School, Worcester; Harvard Medical School, Boston, Massachusetts; University of Maryland School of Nursing, Baltimore; University of Maryland School of Medicine, Baltimore; and Children's National Medical Center, Washington, DC.

The research was supported by the National Institutes of Health (NIH) and the

Muscular Dystrophy Association. This work was also made possible by the National Center for Research Resources (NCRR), a component of the NIH, and the NIH Roadmap for Medical Research. [FSHWatch](#)

Reference

Zhang Y, King OD, Rahimov F, Jones TI, Ward CW, Kerr JP, Liu N, Emerson CP Jr, Kunkel LM, Partridge TA, Wagner KR. Human skeletal muscle xenograft as a new preclinical model for muscle disorders. *Human Molecular Genetics*. February 7, 2014.



NEWS AND EVENTS

FUNDRAISING HOW-TO WEBINARS

Have you thought about organizing a fundraising event but don't know how to get off the ground? Check out our series of fundraising "master class" webinars, available for free on the Web. We have held two to date, on auctions with Terry of Colella from Friends of FSH Research on March 14, and on benefit concerts with Judy Seslowe and Beth Johnston on April 4. Keep an eye out for our email invitations. The recorded webinars are also posted on the FSH Society's YouTube channel. Special thanks to Tony Teel for editing the videos.



NOT GETTING OUR EMAILS?

The FSH Society sends out email notices at least once or twice a month. If you have not been receiving them, you are either not on our distribution list, or our emails are getting trapped. Please check your spam folder, and if that is not the problem, contact us at info@fshsociety.org and ask to be added to the list. We don't want you to miss out!

ERRATUM

In the Winter 2014 issue of the *FSH Watch*, we inadvertently left out Amy Bekier's name from our photo feature on the Celebrity Walk 'n' Roll. Amy founded and chairs the Walk. Our most sincere apologies. We also omitted Howard Chabner's name under proofreading and editing. Thank you, Amy and Howard, for your hard work and dedication!

Why Does FSHD Target Muscles?

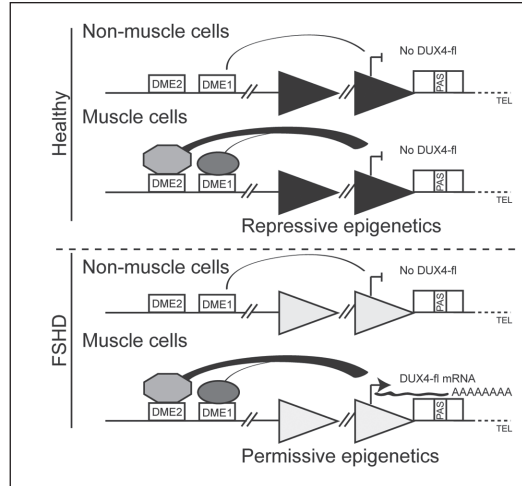
UMass team has a hypothesis

by JUNE KINOSHITA
FSH Society

The genetic mechanism implicated in FSHD—the dysregulation of the D4Z4 region on chromosome 4—is present in every cell of a patient’s body, and yet the disease singles out the skeletal muscles for damage. Why? A University of Massachusetts Medical School (UMMS) study has found a possible explanation: two regulatory elements on chromosome 4 that enhance the expression of the suspected toxic gene DUX4 in muscle cells, but not in other cell types.

The DUX4 gene is widely thought to play an important role in FSHD. So far, two genetic abnormalities have been linked to the disease. FSHD Type 1 is associated with a loss of D4Z4 units on chromosome 4, while FSHD Type 2 is associated with mutations of a gene called SMCHD1 on chromosome 18. Although quite distinct, both genetic abnormalities converge on the same result: They change the D4Z4 region so that the DUX4 gene, which is normally in “lock down” and unexpressed, can become expressed. (More than 95 percent of FSHD patients have FSHD Type 1. The range and severity of symptoms are the same for both types.)

In order for any gene to become expressed, a variety of proteins first are called into play that interact with the gene’s regulatory sequences to trigger the process by which the gene gets “transcribed” into messenger RNA. In a study published in *Molecular and Cellular Biology*, the UMMS team reported that two DNA sequences, which



Schematic showing how DME1 and DME2 have no effect on DUX4-fl protein expression in healthy muscle cells or in FSHD non-muscle cells, but activate DUX4-fl expression only in muscle cells from FSHD patients. Diagram courtesy of Peter Jones.

they named DUX4 myogenic enhancer 1 (DME1) and DUX4 myogenic enhancer 2 (DME2), activate DUX4 expression in skeletal muscle, but not in other cell types.

The finding provides “an explanation for why FSHD pathology is largely restricted to muscle,” says study co-author Takako Jones, PhD.

“Previously, very little was known about the mechanisms controlling DUX4 expression in FSHD,” Jones said. “Gaining a better understanding of the factors and signaling pathways that converge on these enhancers to regulate DUX4 will enable us to more accurately recapitulate FSHD-like pathology in model organisms and pave the way for safer and more specific therapeutic strategies.”

The study was supported by grants from the National Institutes of Health; Association Française contre les Myopathies; the Thoracic Foundation; and the Muscular Dystrophy Association. The paper thanks the FSH Society for outreach to FSHD patients who contributed muscle biopsies, and the Wellstone Muscular Dystrophy Cooperative Research Center for obtaining the biopsies and deriving the original cell cultures used in the study. **FSH Watch**

Reference

Himeda CL, Debarnot C, Homma S, Beermann ML, Miller JB, Jones PL, Jones TL. Myogenic enhancers regulate expression of the facioscapulohumeral muscular dystrophy associated DUX4 gene. *Mol Cell Biol*. 2014 Mar 17. Epub 2014 Mar 17.



INFORMATION AT YOUR FINGERTIPS

The FSH Society’s publications for patients are written and reviewed by teams of experts. We are dedicated to making sure you have accurate, useful information to help improve the quality of your healthcare and daily life. You can download these from our website (<http://www.fshsociety.org>) or request printed copies by contacting us at:

► FSH Society
450 Bedford Street
Lexington, MA 02420
Telephone: 781-301-6060
Email: info@fshsociety.org

FSHD Patient Brochure (in English)
http://www.fshsociety.org/assets/pdf/PatientBrochure_May2009.pdf

FSHD Patient Brochure (in Spanish)
<http://www.fshsociety.org/assets/html/PatientBrochureSpanish.html>

Exercise, Physical Therapy and FSHD
<http://www.fshsociety.org/pages/patHlExer.html>

FSHD: A Guide for Schools
http://www.fshsociety.org/assets/pdf/FSHSociety_FSHDPubs_AGuideForSchools.pdf





Clarice: A Life of Love and Courage

Part two of a family saga

by **TIM GUZIK**
Crescent City, California

In the previous issue of FSH Watch, Tim Guzik shared the story of his family and his four siblings affected by FSHD. This is the second installment of the Guzik family story.

Our parents met while serving our country during World War II. They fell in love, married, and like so many other couples of the Greatest Generation, settled into creating a home and starting a family. Sam and Fran Guzik had learned from the terrible years of the war to face life with courage and that it was important to live purposeful lives in order to honor the sacrifices of the fallen.

Mom and Dad began their family immediately after they were married, and within a few years were blessed with five sons. My parents were devout Catholics and loved each child as a blessing from God. However, my mother wanted a little girl and began a devotion to St. Jude, the patron saint of impossibilities. My mother's prayers were answered and our sister Clarice was born on July 4, 1955.

Clarice was captivating with her auburn hair, dark brown eyes, and sweet disposition. Our mother dressed her in cute little frocks that made her irresistible. Clarice quickly became the "Queen" of the family and center of our attention.

In fact, Clarice didn't walk until she was over two years old because she was held constantly by everyone. Clarice just didn't feel the need to walk when she could be carried everywhere. We assumed it was due to our constant holding, but in retrospect I wonder if her little legs just weren't strong enough due to FSHD.

Ultimately, our parents had 13 children—eight sons and five daughters. Clarice grew into a lovely young woman and was always revered as the "Queen" of the family.

When Clarice entered adolescence her body began to exhibit the signs of FSH muscular dystrophy. The muscles in her face weakened, making facial expressions difficult. Her shoulders slumped forward and her back began to curve. She began to walk with a slight limp and eventually began to thrust her feet forward, struggling with each step.

Her body was going through profound changes that could have had a devastating effect on her self-esteem. But Clarice showed the depth of her character by working hard to always look her best and focus on her abilities rather than her disabilities.

Clarice was the only sister along with three brothers who would be stricken with this insidious disease. She faced the many formidable and progressive challenges of the disease with her characteristic strength and determination.

As a young woman, Clarice loved sew-

ing clothes. She sewed many of her own clothes and worked for several years in a sewing store. Clarice's hair, makeup, and clothes were always impeccable and complemented her natural beauty.

Another passion of Clarice's was cooking and baking. She would make the most amazing and delicious dishes and desserts for our family gatherings. She knew her big brother Tim loved her date-filled cookies and would always make them during the holidays. Clarice adored her family and was at every family event sharing her love.

In her early twenties Clarice met husband Danny, whose occupation was in real estate, and he encouraged her to join him in the business. Clarice studied hard and passed the real estate exam, and so began a long and successful career. Clarice's FSHD was continuing its slow but relentless progression, but she would never allow it to determine what she wanted to accomplish in her life.

After a few years of marriage, Clarice and Danny contemplated having a baby. Clarice knew that carrying a baby would be physically demanding for her, and there was a possibility the child might be born with FSHD. Clarice decided she could handle pregnancy and the rigors of motherhood, and gave birth to little Valerie. Clarice was a strong, loving, and dedicated mother. She adored her baby girl and raised her to be strong and independent.

Clarice's marriage to Danny eventually ended, but Danny continued to play a supportive role in Clarice's and Valerie's lives. Eventually, Clarice found a loving partner named Jim and had a beautiful relationship for over 22 years.

Clarice continued to work in San Clemente real estate until her FSHD made it nearly impossible for her to show properties. She was very successful and was well known among her clients and the citizens of San Clemente. During most weekends Clarice would use her electric wheelchair to browse the downtown stores, where she was warmly greeted by everybody in the town.

Deciding that she needed and wanted to continue to work, Clarice went back to college and earned a degree from the University of California, Irvine in Social Ecology. Her goal was to become an advocate for the disabled, and she was hired as a social worker for the Dayle McIntosh Center.

Clarice loved her work and was an inspiration for her clients. Due to her advanced illness, she labored for several hours each morning to get ready for work, and then she would drive her wheelchair into her van and



Clarice Ventura at the Celebrity Walk 'n' Roll.



The First Phil's Jam for FSH

Nashville singer-songwriters give voice to #fshd

by PHIL BENNETT
Nashville, Tennessee

drive herself to her office. She never gave up, she never complained, and she always inspired everyone she touched.

Clarice loved people with challenges, and was always contributing to charities and attending events that raised money to help find cures for diseases. When she was finally forced to retire due to her FSHD, she immediately began to work on an FSH Society annual event in Irvine called Celebrity Walk 'n' Roll to raise money to help find a cure. The event raised over \$50,000 and was a tremendous success.

Clarice never gave up hope that a cure would be found—if not for her and her family, then for all those families with loved ones affected by this disease.

A few months before Clarice died, she wanted to send her housekeeper and family to Disneyland. She contacted her nephew Nathan, who works for ABC Studios, and with his help purchased tickets for the entire family.

Clarice had a soul that was filled with compassion, love, understanding, and empathy. She was never intimidated by her physical limitations and always saw problems as challenges that had a solution if she tried hard enough.

One time during a family camping reunion in Yosemite, the family wanted to swim in the Merced River, which flows through the campground. The shore was filled with large boulders and rocks, making it nearly impossible for Clarice to join her family at the river. Clarice was determined to enjoy the river with everybody else but couldn't drive her wheelchair over the rocks. So she summoned her three strongest brothers and instructed them that they were to carry her over the dangerous rocks. She completely trusted that her three brothers would carefully and lovingly carry her to be with her beloved family.

Clarice was raised with a strong sense of faith, but in her later years, her faith grew and so, too, her desire to share that faith with others. She read the Bible, attended Bible study classes, and eagerly shared her love of God with others. Because of her strong faith, several family members came to know the love of God that radiated from Clarice's spirit.

Clarice demonstrated her strength and deep faith during her final hours in the hospital. Her FSHD had weakened her lungs to the point that she could no longer breathe on her own. She was placed on a ventilator and would never again be able to live her life without being tethered to a machine. Clarice had a powerful belief that in heaven she would be free to watch over her family until we joined her beautiful spirit.

Clarice made the decision to turn off the ventilator and allow God's will to determine her future. She was surrounded by her family as she slowly took her last breath and entered into the loving arms of God. No one in our family will ever forget her courage, her faith, and the strength she demonstrated to us in life and in death. FSHWatch

The first Phil's Jam for FSH was held in February 2014 at the Wild Wing Cafe in Franklin, Tennessee. My very first fundraiser featured some of the best singer-songwriters in the Nashville area. There was also a silent auction, food, and drink specials. Wild Wing Cafe donated a portion of their sales to the FSH Society as well.

The night started with Destinee Quinn, finalist on the NBC show *The Voice*, and up-and-coming songwriter and artist AJ Engstrom. Special guest Bernie Nelson performed a song he wrote, "Daddy Never Was the Cadillac Kind," which was recorded by Confederate Railroad in 1994 and became their first Top 10 single.

Joel Shewmake entertained the crowd with several of his songs, many of which have been recorded by country music artists including Brad Paisley, Craig Campbell, and Trace Adkins. Kirsty Lee Akers energized the crowd with several original songs. The Anna Johnson Band gave a stellar performance by sharing several songs off their CD *Here*.

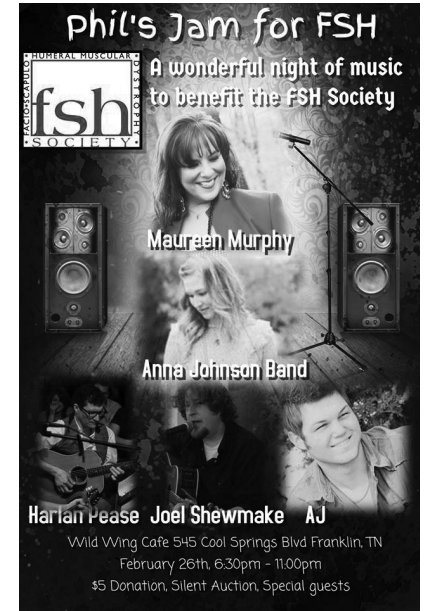
After Anna's band performed, I told my own story and shared a song I wrote inspired by my challenges with FSHD called, "When I Dream." The night ended with R&B singer Maureen Murphy, who blew us away with her amazing vocals.

We sold Phil's Jam for FSH T-shirts and bracelets, which proved quite popular.

One of the highlights of the night was a Twitter challenge. An anonymous donor pledged to donate \$1 to the FSH Society for every tweet with #fshd as the hashtag, up to \$500. This was very exciting as the crowd got quite involved in raising awareness and funds. Over 640 tweets and Facebook posts were shared in less than 24 hours, the most ever for FSHD, it's safe to say. Thank you to this anonymous donor!

Thank you also to Wild Wing Cafe for donating a portion of your sales to the FSH Society. Also, thank you to our first corporate sponsor, KC Phone Guys.

The first Phil's Jam for FSH was such a success that Wild Wing Cafe has offered to host a second Phil's Jam, which is scheduled for Sunday, July 13, 2014. FSHWatch



An Awesome Experience

Getting sliced and diced in an MRI machine

by **JIM FOX**
Bellingham, Washington

In my retirement, I've been volunteering for any trials or research that I can find. There's the University of Rochester FSHD Registry that collects annual data from hundreds of patients and links volunteers with researchers who are seeking research subjects. I've volunteered for several questionnaire surveys, including one that tracks annual symptom progression, which has been going on now for over a decade or so.

We also have a Center of Excellence at the Kennedy Krieger Institute in Baltimore, where FSHD families have donated tissue samples (blood and muscle biopsies) and volunteered for magnetic resonance imaging (MRI) studies. The MRI studies are for both basic research into developing diagnostic techniques as well as for exploring how genetics is expressed in muscle damage and dystrophy. Other exciting studies are in process.

I recommend volunteering as one way we "patients" can help solve the FSHD disorder, leading toward either treatments or a cure. Fundraising and political advocacy are other important ways to contribute.

Recently, after some dynamometer strength tests, I had my second MRI scan. Some folks dislike the experience of being confined in the massive MRI machines, but I find it fascinating and actually enjoy the meditative space odyssey "trip." And knowing that it is state of the art just makes participation better. The leading-edge technology is just too cool! It's awesomely incredible what the resulting MRI scans can reveal.

Mine was an hour or two full-body scan using several scanning (and later, post-processing) protocols. I was strapped tightly onto a sled to prevent movement and surrounded by a cage of white plastic-frame sensors. The sled glided back and forth within the tube-like passage in the massive upended doughnut-shaped MRI machine.

As the sled progressed back and forth, varying magnetic intensity pulses vibrated my molecules (I felt little or nothing), sending radio-like pulse signals to the sensing coils surrounding my body. Different molecules (in bone, muscle, fat, water, etc.) flip and unflip, radiating energy at different frequencies based on their chemical properties. The sensing coils and processing software locate these emissions in three-dimensional space, building a model of my body that can be viewed and studied as various cross-sectional "slices" and "voxel" areas or volumes.

After the scan, I was fortunate to see some of these slice-like internal pictorial results from my toes to the top of my head, or from my left to right side, or front to back.

Post-processing software can display different tissue material (frequencies) as black, dark gray, or white. My leg muscles looked exactly like slices of ham, the solid white bone surrounded by



Magnetic resonance imaging peers into the whole body.

dark meat, marbled with streaks or zones of gray-white gristle and fat. Some muscles look fine, while others are missing or turned to fat, or water infused, indicating possible inflammation.

All the body organs are there, identifiable by shape and position. Using different filters, different tissue properties can be enhanced. We found evidence of some previous medical procedures deep inside me.

It's immediately obvious to me what's generally happening, invisible inside my otherwise impermeable skin surface. My left and right muscles differ, with muscle mass missing or replaced with other, inert tissue. However, the real benefit may be the power of MRI to "see" and statistically quantify the different materials evolving as good muscles become water infused (edema) and deteriorate into fat over time.

All in all I loved it, the incredible experience of my total body being examined, every living molecule vibrated, flipped, and responding, all surveyed with incredibly detailed sensory and data-processing routines, producing awesome images that can be later

studied and measured.

Truly state-of-the-art, leading-edge stuff. Life is good, and this damnable FSHD can also be fascinating. Thank you to our dedicated researchers and research sponsors! [FSHWatch](#)

Link:

Volunteers With FSHD and Volunteers Without FSHD Are Needed!
http://www.fshsociety.org/assets/pdf/WagnerLeungMRI_Recruitment_FSHSocietyGlossy.pdf

Get Involved With Research!

FSHD-affected and FSHD-unaffected volunteers needed for research study using MRI and magnetic resonance spectroscopy (MRS) imaging in skeletal muscle at KKI, Baltimore, Maryland.



Six New Research Grants Awarded

Innovative research propels the ongoing search for FSHD treatments and cures

by **DANIEL PAUL PEREZ**
FSH Society

Earlier this year, the FSH Society awarded six grants totaling \$609,565 to new research projects. Through these studies, the Society's fellowship program aims to gain insights and achieve significant milestones in understanding FSHD, one of the most prevalent types of muscular dystrophy.

The Society received so many proposals for its August 2013 round that its Scientific Advisory Board had to schedule two extra sessions to review them all. Thanks to the strong growth in donations from FSH Society members and benefactors, the Society was able to award larger grants in 2014, which ensure that these important projects can move forward more rapidly and robustly.

The following projects were awarded funding in February 2014:

► **INVESTIGATING EFFECTS OF PARP1 INHIBITORS IN DUX4 EXPRESSION (\$89,267)**

Yi-Wen Chen, DVM, PhD, George Washington University and Children's National Medical Center (Washington, DC)

Summary: A mysterious protein called DUX4 is believed to cause FSHD. The findings of the study will provide insights into the involvement of PARP1 in regulating the amount of DUX4 in FSHD, and will have a direct impact on developing therapeutics for FSHD.

► **GENE SURGERY USING TALEN TECHNOLOGY: A THERAPY FOR FSHD (\$117,500)**

Julie Dumonceaux, PhD, Institut de Myologie, University of Paris, U974 INSERM (Paris, France)

Summary: The approach proposed in this study—unlike other therapeutic strategies under investigation for FSHD—does not require repeated long-term administration of treatment. The benefits of this as a clinical therapy include lower cost and reduced toxicological and immunological risk. Moreover, this approach would be useful for all FSHD cases, regardless of the precise mutation or contraction involved.

► **PROTEIN CHEMISTRY AND PROTEIN-PROTEIN INTERACTIONS OF DUX4 (\$70,000)**

Jocelyn Eidahl, PhD, The Research Institute at Nationwide Children's Hospital (Columbus, OH)

Summary: DUX4 has been identified as a potential cause for FSHD, but the mechanisms by which DUX4 contributes to FSHD pathologies is unclear. The study's hypothesis is that the DUX4 transcription factor is involved in protein-protein interactions that influence its ability to induce toxicity in muscle cells and ultimately contribute to FSHD. The study examines the functional significance of protein-protein interactions of DUX4 that are critical for DUX4 toxicity.

► **EXPLOITING GENOME EDITING TECHNOLOGY TO MODIFY AND REGULATE THE FSHD DISEASE LOCUS (\$125,000)**

Funding for this project was made possible in part by a gift from the FSHD Canada Foundation.

Michael Kyba, PhD, Lillehei Heart Institute, University of Minnesota (Minneapolis, MN)

Summary: Recent discoveries of DNA-binding factors have opened up tremendous new possibilities in genome editing. Through the grant, this study will take advantage of and leverage an existing research program in genome editing of FSHD induced pluripotent stem cells (iPSCs), and will provide the field with valuable new tools to study the pathogenesis of FSHD, and develop cell therapies based on corrected, isogenic iPSCs.



► **MICRODIALYSIS FOR THE STUDY OF INFLAMMATORY FEATURES IN FSHD (\$70,000)**

Giorgio Tasca, MD, Institute of Neurology, Catholic University School of Medicine (Rome, Italy)

Summary: The study will implement a technique that has never been applied to the study of skeletal muscle and will provide a better understanding of the role of the inflammatory process in the disease, the identification of biomarkers of disease activity at the single muscle level, and the acquisition of information useful for the development of a targeted anti-inflammatory therapy. In the future, the new technique could be used for molecular monitoring and eventually drug administration in neuromuscular disorders.

► **DYNAMIC MAPPING OF PERTURBED SIGNALING UNDERLYING FSHD (\$137,798)**

Peter S. Zammit, PhD, King's College (London, UK)

Summary: This study will map changes in cell signaling in FSHD that could identify targets for treatments to help augment muscle repair in FSHD, reversing muscle weakness and wasting. The research's ultimate aim is to gain knowledge of muscle regeneration in FSHD to inform the design of therapies.

These new studies represent a crucial step in the ongoing development of FSHD knowledge, treatments, and cures. We are thrilled to award the grants to such innovative research endeavors, which bring us closer to finding treatments—and medical breakthroughs—for FSHD.

The FSH Society fellowship program allows research programs to develop, prove successful, and ultimately to attract funding from large funding sources such as the US National Institutes of Health (NIH) and large private sources. [FSHWatch](#)



Opera Legend Frederica von Stade to Perform at FSHD Benefit

San Francisco concert and auction planned for July 24


by JUNE KINOSHITA
FSH Society

San Francisco Bay Area audiences will have an opportunity to enjoy a stellar concert to benefit the FSH Society this July. Featuring Frederica von Stade, one of the world's most celebrated mezzo sopranos, and Steven Blier, the acclaimed pianist, the concert will take place at Yoshi's at 1330 Fillmore Street, San Francisco, on Thursday, July 24, 2014.

Joining them for a program drawn from the Great American Songbook, French popular songs, Latin American music, and a touch of Kurt Weill will be rising stars in the San Francisco Opera Merola Opera Program. These are "songs that have continually fueled my imagination and my heart," says Blier.

Yoshi's, a 400-seat, state-of-the-art live music club and Michelin-rated Japanese restaurant, will provide a memorable and festive setting for the concert—the first major benefit for the FSH Society ever held in the Bay Area. The concert will be preceded by a silent and live auction.

Event tickets include a light dinner, auction, and concert and are priced at \$125. The VIP package includes, in addition, an auction preview, exclusive VIP reception, meet and greet with the artists, and a gift; it is priced at \$250. Tickets for just the auction and concert are \$50. Please visit the FSH Society website to reserve your tickets.

Event co-chairs Joyce Hakansson and Ann O'Leary would welcome inquiries about joining the host committee, auction item donations, event sponsorships, and volunteering. Please contact them at annoleary@yahoo.com or joyce@hakansson.com if you would like to help, or have connections or resources that could be of benefit. June Kinoshita can be contacted at june.kinoshita@fshsociety.org. Thank you! 



Frederica von Stade

GET SOCIAL!

Join our online communities to get news, ask questions, and seek advice and support from fellow FSHD patients and family members. The FSH Society Yahoo! Groups forum has tens of thousands of searchable posts. Bookmark these pages and come back often. To find the FSH Society Facebook page and Yahoo! Groups, go to these sites and search for "FSH Society." If privacy is a concern, you can use your account privacy settings to limit who can see your posts. You can also follow us on Twitter @FSHSociety.

MATCHING GIFTS AND OTHER WORKPLACE GIVING

Many employers offer workers options for directing the company's funds to a charitable organization of their choice. When this opportunity is available to you, please consider how your workplace might make a gift to the FSH Society. This is a great way to double, triple, or even quadruple your gift!

CHARITY NAVIGATOR TOP PERFORMER

The FSH Society has been awarded its sixth consecutive Four-Star rating by one of the nation's leading charity watchdog organizations, Charity Navigator, and was named one of America's 10 Charities Worth Watching. Charity Navigator's Four-Star Award—its highest—indicates that the FSH Society consistently executes its mission in a fiscally responsible way and outperforms most other charities in the United States. www.charitynavigator.org



RAZOO ONLINE FUNDRAISING

Razoo provides an easy way for you to create an online campaign. Your donors will enjoy the convenience of giving online and knowing that their gifts will go directly to the FSH Society. Razoo has built-in social media sharing, so you and your friends can help spread the word over Facebook, Twitter, and other social media. <http://www.razoo.com/story/Facioscapulohumeral-Society>



2014 Events: Save the Date!

For details and registration, please check the FSH Society website at www.fshsociety.org

May 10: Mid-Atlantic Patient Network Meeting Garden Party Kennedy Krieger Institute, Baltimore, Maryland

May 17: Patient Support Group Meeting, co-hosted with Friends of FSH Research. Fred Hutchinson Research Center, Seattle, Washington

June 14: Third Annual Spicerfest benefit concert. Murphy's in Memphis, Tennessee

July 5: Second Annual Lakeside Celebration. Hickory Corners, Michigan

July 13: Phil's Second 2014 Jam for FSH. Wild Wing Cafe, Franklin, Tennessee

July 24: Songs in the Key of Steven Blier, a benefit auction and concert with pianist Steven Blier and opera superstar Frederica von Stade. Yoshi's, San Francisco, California

August 16-17: FSH Society FSHD Connect Conference. Boston, Massachusetts

September 6: Mid-Atlantic Patient Network Meeting. Kennedy Krieger Institute, Baltimore, Maryland

September 13: Fifth Annual Fulmer Family Dinner. Sharon Baptist Church, McDonough, Georgia

September 29: A Festive Evening of Song, a benefit auction and concert with Steven Blier and Sylvia McNair. Tappan Hill, Tarrytown, New York

October 3: Third Annual Hustle4Muscle Golf Tournament. Abilene, Texas

October 12: Fifth Annual Celebrity Walk 'n' Roll. Irvine, California

November 15: 16th Annual Friends Supporting Hope Fundraiser. Boston.