

FSH Watch



CONNECTING THE COMMUNITY OF PATIENTS, FAMILIES, CLINICIANS AND INVESTIGATORS



RESEARCH

A long-awaited mouse offers a new animal model for FSH muscular dystrophy

by JUNE KINOSHITA, FSH SOCIETY, EXECUTIVE DIRECTOR
Lexington, Massachusetts

It's a truism of medical research that humans are cheaper and better to study than animals. People don't require sterile cages, gourmet chow or 24/7 monitoring by a battalion of technicians. And because of species differences, diseases and cures in animals often don't accurately portray the human condition. Still, animals remain invaluable because they can be manipulated and probed in ways that human volunteers (and ethical boards) might frown upon.

When a disease is unique to humans, creating an animal model presents a particular challenge. Such has been the case with FSHD. Past efforts resulted in mice that exhibited features of the disease, but which required cranking up the expression of genes under conditions that researchers fear were too dissimilar to the human disorder.

Now an international team has published a mouse model that appears more promising. The result of a decade's worth of work, during which scientific understanding of FSHD exploded, the new mouse was published in April in an exhaustive, 58-page article in *PLoS Genetics*. "We hope that in the near future these mouse models will

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LIVING WITH FSHD

The Centers for Disease Control Spotlights FSH Muscular Dystrophy

FSH Society member Kevin Kirby speaks about living with FSHD in a Web feature from the U.S. Centers for Disease Control. The Atlanta resident wants to help others facing life with this debilitating muscle disease.

by JUNE KINOSHITA, FSH SOCIETY, EXECUTIVE DIRECTOR
Lexington, Massachusetts

At the age of 28, Kevin Kirby was diagnosed with FSHD. A former distance swimmer and three-time Junior Olympian, Kirby can no longer swim laps, so two summers ago he decided to put a swimming pool to a different use, as a party venue to raise funds for the FSH Society.

FSHD affects an estimated 500,000 people worldwide and is one of the most common muscular dystrophies. Between one and two percent of the general population carries a genetic risk factor linked to FSHD. But while the public is familiar with Duchenne muscular dystrophy, a devastating disease affecting boys, few realize that muscular dystrophy is also a disabling and costly disease among adults.

To ensure that FSHD doesn't get neglected, in 2001 the FSH Society co-authored the Muscular Dystrophy CARE Act, which mandates the Centers for Disease Control (CDC) to work on FSHD. Thanks to this impetus, the CDC decided to spotlight adult dystrophies by featuring Kirby's story on its website this past February.

Earlier this year, Kirby went on disability. But that did not slow him down. He keeps himself busy doing volunteer work for the Humane Society and AIDS vaccine



Kevin Kirby

Photo courtesy of the Centers for Disease Control.

development. And he wants to help patients newly diagnosed with FSHD.

"It's overwhelming to learn you have a disease you can't even pronounce," he says. "Social media is helping connect patients and break down the isolation faced by many with FSHD."

Overcoming the need many patients feel to keep their disease private, Kirby decided to become a public advocate to raise awareness about FSHD. In the CDC feature story, Kirby shares how he tried to remain

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FSH Society Among "10 Charities Worth Watching"



Hope for the Future

Dear Friends,

Spring is here! Outside my window at our new digs in Lexington, I can see a family of Canada geese grazing on the banks of a small pond. The parents stand tall, on the lookout for all threats to the olive-green balls of fluff scurrying around them.



This pastoral scene struck me as an apt metaphor for what the FSH Society does. Co-founded by two FSHD patients and governed by a Board of Directors whose members are comprised almost entirely of individuals who are directly affected by FSHD, the FSH Society keeps a vigilant watch over the interests of FSHD patients and researchers everywhere.

Thanks to our good governance, world-class scientific program and members' strong financial support, the FSH Society has been ranked by Charity Navigator as one of America's "Ten Charities Worth Watching." This issue of the *FSH Watch* is filled with examples of the diverse efforts that have helped the Society attain this prestigious recognition.

When the MD-CARE Act first came before the U.S. Congress in 2001, FSH Society President and CEO Dan Perez made sure that FSHD and seven other dystrophies were included among the muscular dystrophies covered by the Act. With the Act up for renewal this year, he made a powerful case not only to renew federal funding for FSHD research, but to increase it.

In this issue, you can read Dan's Congressional testimony about the impact grant funding has had on FSHD research. You will also learn how the MD-CARE Act mandated the Centers for Disease Control (CDC) to work on adult muscular dystrophies, an initiative which the CDC highlighted in its story on FSH Society member Kevin Kirby.

This issue also features some of the results of FSH Society grants – made possible through your generous support – including a new mouse model, tantaliz-

ing findings about a possible link between FSHD and a genetic structure called the telomere, and the FSH Society's support group meeting hosted by patients and scientists at the Kennedy Krieger Institute in Baltimore, which we are live-streaming over the internet so that people everywhere can tune in.

We are delighted by the news that Dr. John Day and his team at Stanford Hospitals have launched a multidisciplinary clinical center for FSHD, bringing together researchers, medical professionals and patients. We are proud to support their efforts. Patients who are not near such a multidisciplinary center should refer to our information on medical professionals who can help, at www.fshsociety.org, click on "For Patients" at top, then click on "Medical Professionals" on the left or go directly to <http://www.fshsociety.org/pages/patMedProf.html>

We would not be witnessing such a beautiful flowering of results, were it not for the hard work of our volunteers. In this issue we celebrate our Board members, Chris Stenmon, who launched the first FSH Society volunteer-driven fundraiser 15 years ago, and Judy Seslowe and Beth Johnston, whose annual benefit concert has become the Society's largest fundraiser of the year. Alongside, we salute four remarkable young women who want to make a difference: Madison Hooge, Kristina McMullin, Jonelle Spicer and Carden Wyckoff. Their energy, passion and leadership should inspire us all and give us great hope for the future.

Please help us continue our work at the FSH Society with a donation and let's honor all of our volunteers' dedication and hard work by supporting their events as generously as you can.

With best wishes,

June Kinoshita
Executive Director

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

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Articles may be edited for space and clarity. Every effort has been made to ensure accuracy in the newsletter. If you wish to correct an error, please write to the above address.

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Unique Fred Hutchinson Cancer Research Center, GlaxoSmithKline partnership will target muscular dystrophy

by **DEAN FORBES, FRED HUTCHINSON CANCER RESEARCH CENTER**
Seattle, Washington (reprinted with permission)

Discoveries led by Dr. Stephen Tapscott about the genetic and disease mechanisms of FSHD have resulted in a partnership between Fred Hutchinson Cancer Research Center in Seattle and GlaxoSmithKline, PLC, to develop therapeutics to treat the disease.

The goal of the new agreement is to develop a small-molecule-based medicine to potentially reverse FSHD by inhibiting the activity of a protein that is incorrectly expressed by the *DUX4* gene in people with the disease. The protein activity is hypothesized to damage muscle cells and lead to progressive muscle weakness and atrophy in FSHD patients.

The genetic and disease mechanisms of FSHD were discovered by an international team of scientists led by Tapscott in a series of findings published between 2010 and early 2012. Tapscott will lead the Fred Hutchinson work in the GSK collaboration.

The partnership with GSK is a first of its kind for Fred Hutchinson, which is also the first U.S.-based institution to sign on with GSK's "Discovery Partnership with Academia" (DPAC) program. GSK launched the program last year to combine the insight and creativity of the academic world with GSK's drug-discovery expertise to turn innovative research into medicines that benefit patients.

Unlike traditional licensing agreements in which the licensee is given full control to develop a discovery, the collaboration will involve GSK and Fred Hutchinson scientists working together to develop, test and hopefully bring to market a clinical treatment.

"GSK has huge expertise in developing agents against protein activity, so our opportunity to work with them is fantastic," Tapscott said.

FSHD affects about one in 7,500 to 14,000 individuals and usually begins in late adolescence. The effects start around the



Dr. Stephen Tapscott

facial and upper-extremity muscles and eventually progress to muscles in the lower extremity. People with more severe FSHD become wheelchair bound and their life spans are often shortened.

The team's discoveries also have implications for developing cancer immunotherapies because researchers also discovered that *DUX4* regulates cancer/testis antigens. Cancer/testis antigens are encoded by genes that are normally expressed only in the human germ line but are also abnormally expressed in various tumor types, including melanoma and carcinomas of the bladder, lung and liver. This knowledge will give researchers a way to manipulate the expression of cancer/testis antigens, potentially opening the opportunity to use these antigens in a cancer vaccine.

In an era of flat federal research funding, this collaboration signals an increasing interest on the part of Fred Hutchinson to develop partnerships that further its lifesaving and innovative research.

"We're looking for more creative academic-industry partnerships like this one between Fred Hutchinson and GSK," said Dr. Ulrich Mueller, vice president of industry relations and technology transfer at Fred Hutchinson.

The National Institute of Neurological Disorders and Stroke, the National Institute of Arthritis and Musculoskeletal and Skin Diseases and Friends of FSH Research, funded Tapscott's research on FSHD, which provides the scientific basis for the collaboration with GSK.

Financial terms of the Fred Hutchinson-GSK partnership were not disclosed. **FSH Watch**

Editor's note: A series of seed grants from the FSH Society supported early work leading to the discoveries about *DUX4*. Because the mechanisms by which *DUX4* contributes to FSHD are still unknown, the high-risk, basic science funded by the FSH Society remains critically important for the future of FSHD drug discovery.

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, or request printed copies by contacting us at:

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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>

PHYSICAL THERAPY & FSHD
<http://www.fshsociety.org/pages/patHIEExer.html>

FSHD: A GUIDE FOR SCHOOLS
http://www.fshsociety.org/assets/pdf/FSHSociety_FSHDPubs_AGuideForSchools.pdf





FSH Society Submits Congressional Testimony

Request for fiscal year 2014, a doubling of the U.S. NIH FSHD research budget to \$12 million. Testimony of Daniel Paul Perez, President & CEO, FSH Society, Inc. before the United States Senate Appropriations Committee Subcommittee on Labor, Health and Human Services, and Education

On the subject of \$12 million FY2014 Appropriations for U.S. DHHS National Institutes of Health (NIH) Research Programs on Facioscapulohumeral Muscular Dystrophy (FSHD)
May 6, 2013

Honorable Chairwoman Mikulski and Ranking Member Harkin, thank you for the opportunity to submit this testimony. I am Daniel Paul Perez, of Bedford, Massachusetts, President and CEO of the FSH Society, Inc. and an individual who has lived with facioscapulohumeral muscular dystrophy (FSHD) for 51 years. For hundreds of thousands of men, women, and children worldwide the major consequence of inheriting this form of muscular dystrophy is a lifelong progressive loss of all skeletal muscles. FSHD is a crippling and life shortening disease. No one is immune. It is both genetically and spontaneously transmitted to children. It can affect multiple generations and entire family constellations.

The National Institutes of Health (NIH) is the principal source of funding of research on Facioscapulohumeral Muscular Dystrophy (FSHD) currently at the \$6 million level. Over many years, this Committee has supported the incremental growth in funding for FSHD research. I am pleased to report that this modest investment has produced huge scientific returns.

1. CONGRESS HAS MADE A MAJOR DIFFERENCE IN MUSCULAR DYSTROPHY

I have testified many times before Congress. When I first testified, we did not know the mechanism of this disease. Now we do. When I first testified, we assumed that FSHD was a rare form of muscular dystrophy. Now we understand it to be one of the most prevalent forms of muscular dystrophy. Congress is responsible for this success, through its sustaining support of the National Institutes of Health (NIH), and the enactment of the Muscular Dystrophy CARE Act. I am testifying in order to document this success and call on Congress to continue the momentum of discovery you have set in motion.

Congress enacted The Muscular Dystrophy Community Assistance, Research and Education Amendments of 2001 (the MD-CARE Act, Public Law 107-84) on December 18, 2001. It was reauthorized in 2008 and new efforts are underway to reauthorize the MD-CARE Act, as it will expire in 2013. We are hopeful that this reauthorization bill will receive the same overwhelming bi-partisan support enjoyed in earlier enactments.

2. QUANTUM LEAPS IN OUR UNDERSTANDING OF FSHD HAVE OCCURRED IN PAST THREE YEARS

The past three years have seen remarkable contributions made by researchers funded by NIH.

- On August 19, 2010, American and Dutch researchers published a paper which dramatically expanded our understanding of the mechanism of FSHD.¹ A front page story in the New York Times quoted the NIH Director Dr. Francis Collins saying, "If we were thinking of a collection of the genome's greatest hits, this would go on the list."²



- Two months later, another paper was published that made a second critical advance in determining the cause of FSHD.³ The research shows that FSHD is caused by the inefficient suppression of a gene that may be normally expressed only in early development.
- On January 17, 2012, an international team of researchers based out of Seattle discovered a gene called *DUX4* required to develop chromosome 4-linked FSHD.⁴
- Six months later, another high profile paper produced by the NIH funded University of Massachusetts Senator Paul D. Wellstone Cooperative Research Center for FSHD, used sufficiently "powered" large collections of genetically matched FSHD cell lines generated by the NIH center that are both unique in scope and shared with all researchers worldwide, to improve on the Seattle group's finding by postulating that *DUX4*-fl expression is necessary but not sufficient by itself for FSHD muscle pathology.⁵ This work was also supported by a NIH cooperative research center grant mandated by the MD-CARE Act.
- On July 13, 2012, a team of international researchers from the, United States, Netherlands and France identified mutations in a gene causing 80% of another form of FSHD. This paper furthers our understanding of the molecular pathophysiology of FSHD. This work too was supported in part by a program project grant from NIH.⁶
- On April 4, 2013, an international team published a mouse model that appears more promising than previous models of FSHD. This mouse model was the result of a decade's worth of work, during which scientific understanding of FSHD exploded. "We hope that in the near future these mouse models will serve an important purpose in drug development programs for FSHD," remarked senior author Silvere van der Maarel of Leiden University in the Netherlands. The herculean project

was initiated in 2003, by the FSH Society's Marjorie Bronfman Fellowship grant. The patient-driven charity was seeking a definitive mouse model based on a genetic unit called D4Z4. Normally, people have ten or more of these units, repeated one after the other near the tip of chromosome 4. The majority of FSHD patients, in contrast, have fewer than ten D4Z4 units. The newly published mouse model contains 2.5 copies of the D4Z4 unit, a truncated number comparable to that seen in human FSHD patients. The D4Z4 unit contains the gene called *DUX4*, which is toxic to muscle cells.⁷ This work was also supported by NIH grants.

I am proud to say that many of these researchers have started their efforts in FSHD with seed funding from the FSH Society and have received continued support from the FSH Society, the National Institutes of Health, and the Muscular Dystrophy Association and other partners. This shows the power of the collaboration among funders, patient groups and researchers to advance the search for cures and treatments.

3. REMARKABLE PROGRESS IN FSHD RESEARCH AND THE NEED TO KEEP MOVING FORWARD

Given the recent developments, there is a need to ramp up the preclinical enterprise and build/organize infrastructure needed to conduct clinical trials. Our immediate priorities should be to confirm the new hypotheses and targets. We need to be prepared for this new era in the science of FSHD, by accelerating efforts in the following five areas:⁸

1. Genetics / epigenetics. There is general acceptance that transcriptional deregulation of D4Z4 is central to FSHD1 and FSHD2. The FSHD2 gene *SMCHD1* explains approximately 80% of FSHD2. There is a need for better understanding of the factors that modulate *DUX4* activity and disease penetrance.

2. FSHD molecular networks. D4Z4 chromatin relaxation on FSHD-permissive chromosome-4 haplotypes leads to activation of downstream molecular networks. In addition to considering *DUX4* as the "target" and downstream targets, the upstream processes and targets – triggering of activation – are equally important. Hence, understanding what *DUX4lf* does as a target and targets up- and down-stream of it are priorities. Detailed studies on these processes are crucial for insight in the molecular mechanisms of FSHD pathogenesis and may contribute to explaining the large intra- and interfamily clinical variability. Importantly such work may lead to intervention (possibly also prevention) targets. Additional FSHD genes and modifiers are still likely to exist. Apart from chromatin modifiers, these include, but are not limited to, *CAPN3* and the *FAT1* gene that was recently suggested to be involved in FSHD.

3. Clinical trial readiness. It is now broadly accepted that deregulation of the expression of D4Z4 / *DUX4* is at the heart of FSHD1 and FSHD2. This finding opens perspectives for intervention along different avenues. Intervention trials are envisaged within the next several years. The FSHD field needs to be prepared for this crucial step. There is an increasing need to improve the translational process. This includes, but is not limited to, the need for consensus on data capture and storage, overcoming national and international barriers, definition of natural history, identification of (meaningful) and sensitive outcome measures, biomarkers, and meaningful functional measures. There is a need to work more closely with the FDA to help define acceptable measures for trials.

4. Model systems. There was already a good set of cellular models, based on different pathogenic (candidate gene) hypotheses. This was further expanded during the last year. The phenotypes are very diverse and often difficult to compare with the human FSHD phenotype. Many basic questions remain unanswered and dearly need to be answered for further translational studies: when and

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► FSHD Research Dollars & FSHD as a Percentage of Total NIH Muscular Dystrophy Funding

Sources: NIH/OD Budget Office & NIH OCPL & NIH RCDC RePORT

(e = estimate; as FY2012 actuals were not available on-line as of March 12, 2013) In millions

	2000	2001	2002	2003	2004	2005	2006	2007	2008	2009	2010	2011	2012e
ALL MD*	\$12.6	\$21	\$27.6	\$39.1	\$38.7	\$39.5	\$39.9	\$47.2	\$56	\$83	\$86	\$75	\$75
FSHD*	\$0.4	\$0.5	\$1.3	\$1.5	\$2.2	\$2.0	\$1.7	\$3	\$3	\$5	\$6	\$6	\$6
FSHD (% of MD)	3%	2%	5%	4%	6%	5%	4%	5%	4%	6%	7%	8%	8%

MD = muscular dystrophy * In millions of dollars

“You Can’t Be Afraid.”

The FSH Society salutes Board members Judy Seslowe and Beth Johnston

by JUNE KINOSHITA, FSH SOCIETY, EXECUTIVE DIRECTOR
Lexington, Massachusetts

This September 30th, the grounds of the New York Botanical Gardens will begin to glow with the first russet tinge of autumn foliage. Fall-blooming asters and anemones will erupt amidst plumes of tall grass. And as night descends, if you are lucky enough to be there, you will feel the air resonate with sublime music: fluid melodies tumbling forth from a piano and frolicking with a rich, lyrical soprano voice.

The two artists making this wonderful music are among the brightest stars of New York’s cultural firmament: revered pianist Steven Blier and singer-actress Judy Kaye, one of Broadway’s leading ladies. How is it that a pianist whom *New York Times* columnist Joe Nocera called “one of the most extraordinary people I know”, and a two-time Tony award winner, are joining together to perform a concert to benefit the FSH Society? By evening’s end, they hope to have exceeded the \$244,000 raised at last year’s concert.

“It all started over dinner,” says Judy Seslowe, who co-chairs the concert. The FSH Society’s then-Executive Director Nancy Van Zant had encouraged restauranteurs Abigail and Bob Kirsch to host a group of FSHD patients and families at their venue at the botanical gardens. It was there that Seslowe met Beth Johnston, whose husband Jeff had recently been diagnosed with FSHD.

Seslowe had attended an FSH Society benefit concert several years earlier featuring Steven Blier, who has FSHD, and violinist Hanna Lachert. “I wanted to revive the concert idea but wasn’t getting anywhere,” recalls Seslowe. “At that dinner, I asked if anyone wanted to help me, and Beth was enthusiastic.” Seslowe would not have spoken up, had Van Zant not urged her on. “I was so shy!!”

Fast forward several years, and it’s hard to believe Seslowe wasn’t born to be an impresario. Right around Ground Hog day, with the snow still on the ground, she and Johnston rally their committee members, drawn from family, friends and FSHD patients and advocates, and start sketching a battle plan.

Steven Blier performs every year, but who will share the stage with him? E-mails



Beth Johnston and Judy Seslowe

and phone calls fly back and forth until that cornerstone is cemented in place. “Steven knows so many people in New York City, and has been able to produce talent appropriate to this crowd, to make a fun, light evening,” Seslowe praises. “He’s the consummate entertainer, and so committed.” Last year’s concert featured another major Broadway star, Kelli O’Hara.

Seslowe and Johnston say they could never pull off the concert without their families and friends. The committee members gather a thousand names, making sure all the addresses are up to date. They lay out a timeline, draw up a budget, figure out sponsorship and ticket prices, draft a save-the-date letter, create invitations, negotiate contracts for the venue, catering, piano rentals, audio and visual equipment, staffing... the list goes on and on.

Committee members fan out to gather auction items, flower arrangements, decorations. There are endless appeals to family and friends to “please come this year” – and to push their social network to attend or send a contribution.

Seslowe’s family pitches in to help in ways large and larger. Her husband Ken, daughter Emily, daughter-in-law Jamie and friends Sheila Cohen and Sue-ann Friedman assemble the beautifully designed and printed Journal to honor the event sponsors. Other family members send in

generous gifts to help sponsor the event.

Last year, Seslowe’s sister Susan Egert and brother-in-law Bill Milling wrote, produced and filmed a professional video about FSHD and the work of the FSH Society (which you can view on the FSH Society’s homepage). The video, which features Steven Blier, Kelli O’Hara, Seslowe’s brother Bill, Beth Johnston’s daughters, and other patients, was debuted at last year’s concert. Milling’s American Movie Company is creating a new video for this year’s concert, calling in dozens of professional favors to complete the shooting and editing.

Seslowe has special praise for her co-chair. “Beth is a star. She’s a tireless worker, extremely organized and talented.” Johnston laughs about this. “We make such a good team. Judy is so good at talking to people and rounding them up, and I run the operations.”

Johnston admits that she’s “a big keeper of lists. I keep everything from every year. I never re-invent the wheel. I take notes at every meeting. We always have a post-mortem after each concert and talk about what worked, what didn’t. That’s critical. You have to build on what you’ve learned. I’m also a spreadsheet queen!”

It has been “a learning experience,” Seslowe says understatedly. “I was amazed at the friends who stepped forward. I never

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FSH SOCIETY SUBMITS CONGRESSIONAL TESTIMONY

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where is *DUX4* expressed in skeletal muscle and what regulates *DUX4* activity. It was recognized that there still exists a gap in our knowledge linking the basic genetic and molecular findings with the observed muscle pathology. The University of Massachusetts NIH Sen. Wellstone center and the University of Rochester continue to generate human cellular resources. These resources continuously deserve attention and need to be replenished. Recent progress in ES-cell technology, including iPS lines, allows for inter-group distribution and dedicated molecular (epi)genetic studies.

5. Sharing. Timely sharing of information and resources remains a critical contributor to the progress in the field. Sharing of resources other information remains a priority (e.g. protocols, guide to FSHD muscle pathology, etc.).

We would be pleased to provide the Committee with detailed information on each of these areas. The pace of discovery and numbers of experts in the field of biological science and clinical medicine working on FSHD are rapidly expanding. Many leading experts are now turning to work on FSHD not only because it is one of the most complicated and challenging problems seen in science, but because it represents the potential for great discoveries, insights into stem cells and transcriptional processes and new ways of treating human disease.

4. NIH FUNDING FOR MUSCULAR DYSTROPHY

Mr. Chairman, these major advances in scientific understanding and epidemiological surveillance are not free. They come at a cost. Since Congress passed the MD-CARE Act, research funding at NIH for muscular dystrophy has increased 4-fold. While FSHD research funding has increased 12-fold during this period, the level of funding is still exceedingly modest.

Despite the great success of the past two and a half years in the science of FSHD brought about by Congress, we are concerned that the budget cuts required by the sequester are coming at a time when many of the FSHD research projects are ending. It is likely that new research projects will not be funded or existing programs will not be renewed. This is a perfect storm that could have devastating effects on FSHD research efforts. I served on the federal advisory committee MDCC for nine years until 2011. We have conveyed to the Executive Secretary of the MDCC our grave concern that the current portfolio of research on FSHD has a disproportionate number of FSHD grants near the end or in the last year of their grant cycles. While most are

competitively renewable this occurrence could not have happened at a worse time with sequestration making meat-axe cuts across all federal agencies.

We request for FY2014, a doubling of the facioscapulohumeral muscular dystrophy (FSHD) research budget to \$12 million dollars. This will allow an expansion of the U.S. DHHS NIH Senator Paul D. Wellstone Muscular Dystrophy Cooperative Research Centers, an increase in research awards, expansion of post-doctoral and clinical training fellowships, and a dedicated center to design and conduct clinical trials on FSHD.

We are aware of the great pressures on the federal budget, but cutting the NIH budget and research funding for FSHD at this time would be the wrong decision. We have come so far with such modest funding. This is not the time to lessen our endeavor. This is the time to fully and expeditiously exploit the advances for which the American taxpayer has paid. Thank you for this opportunity to testify before your committee. **FSH Watch**

Footnotes:

- ¹ Lemmers, RJ, et al, A Unifying Genetic Model for Facioscapulohumeral Muscular Dystrophy *Science* 24 September 2010: Vol. 329 no. 5999 pp. 1650-1653
- ² Kolata, G., Reanimated 'Junk' DNA Is Found to Cause Disease. *New York Times*, Science. Published online: August 19, 2010 <http://www.nytimes.com/2010/08/20/science/20gene.html>
- ³ Snider, L., Geng, L.N., Lemmers, R.J., Kyba, M., Ware, C.B., Nelson, A.M., Tawil, R., Filipkova, G.N., van der Maarel, S.M., Tapscott, S.J., and Miller, D.G. (2010). Facioscapulohumeral dystrophy: incomplete suppression of a retrotransposed gene. *PLoS Genet.* 6, e1001181
- ⁴ Geng et al., *DUX4* Activates Germline Genes, Retroelements, and Immune Mediators: Implications for Facioscapulohumeral Dystrophy, *Developmental Cell* (2012), doi:10.1016/j.devcel.2011.11.013
- ⁵ Jones TI, et al, Facioscapulohumeral muscular dystrophy family studies of *DUX4* expression: evidence for disease modifiers and a quantitative model of pathogenesis. *Hum Mol Genet.* 2012 Oct 15;21(20):4419-30. Epub 2012 Jul 13.
- ⁶ Lemmers, RJ, et al, Digenic inheritance of an SMCHD1 mutation and an FSHD-permissive D4Z4 allele causes facioscapulohumeral muscular dystrophy type 2. *Nat Genet.* 2012 Dec;44(12):1370-4. doi: 10.1038/ng.2454. Epub 2012 Nov 11.
- ⁷ Krom YD, Thijssen PE, Young JM, den Hamer B, Balog J, et al. (2013) Intrinsic Epigenetic Regulation of the D4Z4 Macrosatellite Repeat in a Transgenic Mouse Model for FSHD. *PLoS Genet* 9(4): e1003415. doi:10.1371/journal.pgen.1003415
- ⁸ 2012 FSH Society *FSHD International Research Consortium*, held November 6, 2012 co-sponsored by DHHS NIH NICHD Boston Biomedical Research Institute Senator Paul D. Wellstone MD CRC for FSHD. To read the expanded summary and recommendations of the group see: <http://www.fshsociety.org/pages/sciConsortium.html>

... from previous page

realized how many devoted friends I had."

"You can't be afraid to do this," says Seslowe. Johnston agrees and recounts that back when they first were dreaming about a concert, "We were afraid to ask Bob Kirsch if he'd donate the space at the Botanical Garden."

"At the end of the day," Johnston says, "I'm always completely amazed at how many people came through for us. You can't get something if you don't ask for it. You can't be

afraid to ask. What stands out for me is the astounding generosity of people."

"A really heartening experience for me was to see that so many people want to help," Seslowe agrees. "We feel we've made a difference. The concert has funded many research grants at this point. We're very encouraged. They're now talking about drugs and clinical trials."

"Five years ago we were nowhere," she notes. "It has been so worthwhile, knowing we have so much support, and that we've made a difference."

Seslowe and Johnston encourage others to take the first step. "It's important to get people with FSHD together, because that's how we started. Invite some people to get together." **FSH Watch**

Editor's note: We thank the Seslowe and Johnston families, their friends, FSH Society Board members, patients and our own hardworking staff for this event's success. Would you like to sponsor this year's "Festive Evening of Song" or join the event committee? Please contact june.kinoshita@fshsociety.org.

Surgical methods for addressing facial weakness in FSHD

by **KOFI BOAHENE, M.D., JOHNS HOPKINS HOSPITAL**
 Baltimore, Maryland

Facial expression is an important part of human communication, allowing us to reflect emotions and project non-verbal cues. Small skeletal muscles that mostly originate and connect with the soft tissues of the face are responsible for animating facial expression. The facial muscles also support the facial skin and fat giving it shape and form.

Blinking, whistling, blowing out candles and drinking from a straw are simple day-to-day actions that depend upon the fine action of the facial muscles. These functions are severely affected when the facial muscles become progressively weak as seen in FSH dystrophy. FSHD patients often complain of changes in their speech. They are unable to generate the intraoral pressure to pronounce plosive sounds such as “P” and “B”. As the facial muscles become weaker, mid-facial fat descends, causing the corners of the mouth to turn downwards and giving a falsely sad appearance.

In my surgical practice, I have been developing new reconstructive techniques for FSHD patients. Muscle tone and movement are two attributes to consider when exploring treatment options for facial weakness in these patients.

Muscles in the body are maintained at a slightly contracted state, which is called the muscle tone. The always-activated state of partial contraction keeps the muscles firm, offers some resistance and maintains form and posture. Facial muscle tone keeps the face from drooping, eyelids from sagging, keeps the lips together and offers some cheek resistance when preparing to blow a candle. When the facial muscles become dystrophic, they lose tone and become flaccid.

Facial tone can be improved through procedures to introduce some degree of form, shape, stiffness and resistance to the flaccid areas of the weakened face. Collagen matrix grafts strategically placed in the lower eyelid can lift the eyelid vertically up and allow better eyelid closure. Fat grafting to the buccal space over the flaccid buccinator muscle introduces some resistance to the cheek, which helps with plosive sounds. Targeted

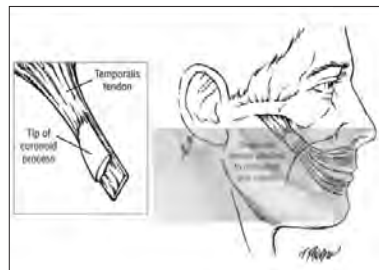
lip augmentation together with muscle placement techniques can be used to correct lip position, shape and closure. In the future, regenerative medicine and tissue engineering research may offer new options for reconstituting dystrophic muscle.

For facial expression and sphincter function around the eyes and mouth, improving tone is not sufficient. One

with another muscle that may also become weak?

One solution is to determine potential donor muscles that are not commonly affected in FSHD. In the head, the temporalis muscle appears to be spared in FSHD. We have successfully transferred the temporalis muscle with techniques uniquely tailored to FSHD patients.

“Facial tone can be improved through procedures to introduce some degree of form, shape, stiffness and resistance to the flaccid areas of the weakened face. Collagen matrix grafts strategically placed in the lower eyelid can lift the eyelid vertically up and allow better eyelid closure. Fat grafting to the buccal space over the flaccid buccinator muscle introduces some resistance to the cheek, which helps with plosive sounds.”



Schematic diagram showing surgical transplantation of the temporalis muscle to restore movement to the mouth.



Collagen matrix graft (white), which can be strategically placed in the lower eyelid to lift up the eyelid and allow better eyelid closure.

needs to restore muscle movement. Although the facial nerves are intact, dystrophic facial muscles do not respond well and animate the face in a very limited manner. Replacing a dystrophic muscle is technically feasible using microsurgical techniques. Microsurgery now allows us to borrow a section of muscle from one part of the body and transplant it to another part to generate a desired effect.

The main limitation in pursuing muscle replacement surgery as treatment for facial FSHD is the progressive nature of the disease and the number of potential donor muscles that can be affected. In other words, why replace a weak muscle

We are investigating other potential donor muscles. The ideal donor muscle should be available for harvest without any significant functional loss and should be spared from progressive dystrophy. Such a muscle can be transferred to the face and reinnervated with the existing facial nerve.

The current options for reanimating the face in FSH dystrophy are promising but should be approached carefully under the direction of a multidisciplinary team that specializes in this disease. FSHWatch

Adapted with permission from Dr. Boahene's website at: <http://www.drboahene.com/Pages/facialparalysis.aspx>



**DR. BOAHENE:
QUESTIONS & ANSWERS
POSED BY PATIENTS WHO ATTENDED
THE BALTIMORE MEETING.**

Q. In order to transplant fat from parts of the body to the face, how is the fat first removed? Does that require invasive surgery and anesthesia?

A. Fat is removed by liposuction using a small 2 millimeter canula. The fat is then gently processed to concentrate healthy fat cells and re-injected to the desired area with a small canula that is less than 1 millimeter wide. The fat can be harvested from various sites including the belly, thigh or arms. Depending on the individual, the procedure could be done under local anesthesia but a level of sedation makes it more comfortable.

Q. How long is the procedure?

A. The whole process takes about one hour.

Q. How long does the beneficial effect usually last?

A. Some of the fat wears off, but the portion that remains is present for several years. When combined with the collagen spacer graft, the effect can last for over five years. A minor touch-up under local anesthesia may be necessary.

Q. What is the recovery time?

A. Some bruising and swelling for about seven to 10 days should be expected.

Q. Is the procedure covered under insurance? If not, what is the estimated cost?

A. This is considered a reconstructive procedure but needs pre-approval from insurance carriers. Response will differ from carrier to carrier.

Q. How soon can one fly after this procedure? If a patient is traveling from a distance to have the procedure done, how long will he or she need to stay in Baltimore before flying home?

A. Patients can physically fly out within 24 hours but we recommend at least a 48 to 72 hour stay in the area for at least one post-procedure check up. International patients may want to wait about five days.

Q. What if the patient is in a weight-loss program? Should the patient wait until after reaching the weight-loss goal before having the procedure done?

A. The number of fat cells transplanted is the key. Those cells do not fluctuate in size significantly with weight loss. Since this is an elective procedure, it is best to perform it when one is in stable health.

For more information, please visit Dr. Kofi Derek Boahene's website with information about surgery for FSHD. (You will need to scroll down the page.) <http://www.drboahene.com/Pages/facialparalysis.aspx>

Why the FSH Society deserves your trust – and gifts

Dear FSH Society Supporters,



In April, the Society received news from Charity Navigator that we had received a 4-star rating for the fifth consecutive year. Ken Berger, President and CEO, Charity Navigator wrote, “only 4% of the charities we rate have received at least 5 consecutive 4 star evaluations, indicating that the FSH Society outperforms most other charities in America!”

As donors, my wife Barbara and I as well as you, have the independent outside opinion that the Society is worthy of the public trust. As a board member with fiduciary responsibility, this recognition speaks to an exceptional job done by our executives, Board members and our Scientific Advisory Board (SAB).

Sixteen of eighteen board members (89%) have personal experience with this debilitating disease. Therefore we all realize that spending money where it counts most, on research and patient support, is critical to finding a treatment and a cure.

Since its inception, the FSH Society has funded grants and commitments totaling over \$4.4 million to research!

Major progress has been made through the funding recommended by our outstanding SAB, which consists of researchers and clinicians renowned in their fields of expertise. Those advances are covered ably by the *FSH Watch* newsletter.

The lifeblood of our organization is donations. Without them we could not provide patient support, fund research, apply for research grants, lobby Congress, maintain liaison with the National Institutes of Health, hold patient meetings and coordinate research with others interested worldwide in the cure and treatment of FSH.

The Society is your leader in improving the lives of hundreds of thousands of patients throughout the world. We are told that the research conducted may also have impact upon other diseases such as cancer and diabetes.

The Society cannot exist without your generosity. Direct gifts of checks, appreciated securities and bequests will help us to continue to help you. Many people have organized fundraising events such as walks, runs, parties, dinners, golf outings and picnics. Some have gotten corporate support as sponsors. Should you wish to organize an event, please contact our Executive Director, June Kinoshita.

The eight members of the development committee, the officers and entire Board are grateful for your past and continued support in our quest for a treatment and cure.

Very proudly yours,

Jim Chin, Sr.

► Festive Evening of Song — September 30

Join us at the New York Botanical Gardens. We would like to thank our committee members below:

- | | | |
|----------------------|---------------------|------------------------|
| Sanford Batkin | Ginny Maxwell | Bob & Abigail Kirsch |
| Steven Blier | Susan Egert-Milling | Jane Rittmaster |
| Jim and Barbara Chin | Maureen Eye | Emily Rosenfeld |
| Sheila Cohen | Sue-ann Friedman | Jamie Seslowe |
| Rosalind Devon | Roslyn Goldstein | Jerry & Marsha Seslowe |
| Jennifer Egert | Beth Johnston | Judy & Ken Seslowe |
| Susan Hecht | June Kinoshita | |

FSH Society Baltimore Patient Researcher Support Group Meeting

by **JIM FOX**
Bellingham, Washington

We had a great FSHD Patient Researcher Support Group Meeting at the Kennedy Krieger Institute (KKI) on the afternoon of Saturday, February 16, 2013, in Baltimore, Maryland. Some 35 attendees came from the mid-Atlantic States, including Pennsylvania, Maryland, Virginia and Washington, D.C.

The meeting was sponsored by an FSH Society grant made possible through a generous donor, and was the result of a lot of work by the FSH Society, KKI staff and volunteers. Billed as “an exploratory event”, it was very successful and appreciated by those attending and involved.

There are four events to be scheduled in Baltimore this year, with possible expansion to other regions in the future. Besides the in-person gathering in Baltimore, live-streaming via the Internet made the meeting available world-wide. Some 48 remote participants logged-in, including one from Australia. We enjoy so much potential through technology these days; all we have to do is harness it.

After introductory remarks from Dr. Kathryn Wagner, director of the Center for Genetic Muscle Disorders at KKI, FSH Society Executive Director June Kinoshita, and meeting coordinators Lilleen Walters and Dr. Genila Bibat, Dr. Kofi Boahene of Johns Hopkins Hospital gave a very interesting talk on surgical techniques that he has developed to address facial muscle weakness in FSHD patients. (See Dr. Boahene’s article on page 8.)

It was fascinating to learn of the many complex muscle groups in the face and throat, and how they affect our eyes, nose, mouth and expressions, along with the techniques Dr. Boahene has developed for improvement or reconstruction. His very cautious, minimally-invasive, progressive and reversible strategies impressed me. So did the advanced surgical techniques for reconstructive surgery which he described as well.

Dr. Boahene divides muscles into two functional groups, for tone and movement (providing shape vs. movement). FSHD affects some muscle groups without affecting others. Remediation tactics range from simply injecting inert or transplanted

“filler”, to propping up or providing a foundation for weak muscles, to adding ties or slings reinforcing weak muscles. Due to rapidly evolving technology, permanent (non-reversible) solutions may not always be best.

Healthy muscles that are not affected in FSHD may be either transplanted, or split and re-directed to aid weak or lost muscles. These must undergo re-training (or ‘rewiring’) of control-nerve impulses. Modern techniques are minimally invasive and carefully designed, taking each individual’s

was good to see a sprinkling of younger and male FSHD folks in attendance. Too often, younger folks and men in particular are reluctant to attend support groups. It is so helpful to be able to share and discuss issues with others who understand our major and often very obscure concerns.

The final session was a discussion of topics for future meetings. These covered the broad range of our experiences, needs and interests. Some are recurring information for new patients, while others are cutting-edge state-of-the-art research findings.




unique FSHD expression into account.

Dr. Boahene is publishing his works. He cautions that any facial reconstruction for FSHD patients should be performed by surgeons very familiar with the special characteristics of FSHD. For example, surgeons must understand which muscles are vulnerable in FSHD, and select for transplantation those muscles that are spared by FSHD. With these caveats in mind, some of our facial deformities and disabilities can indeed be mitigated.

The meeting also had free time with refreshments to simply see old friends and get to know new ones. Attendees included FSHD folks, caregivers and researchers. It

As a participant in various healthcare support groups, I know well how valuable they can be, especially for newly diagnosed patients. We have so much in common and so much to share. And, we are actually making real progress towards mitigating and taming this damnable disorder by working together.

If you have interests in other regional meet-ups, either at a center of excellence or locally, just do it! Our FSH Society recognizes the potential, especially for two-way communications between researchers and patients. With so many people affected by FSHD, working together we can finally make a difference. 

Banking on a Cure

by **CHERYL HOOGE**

Vancouver, Canada

Editor's note: This meeting was funded through an FSH Society grant made possible by a generous donor. We thank Lilleen Walters, who has long been involved with a regional patient group, along with Drs. Kathryn Wagner and Geni Bibat of KKI for their dedication to ensuring the success of the meeting. Special thanks to Don Burke, Frank Kolakowski and Alan Brown for lending valuable technical assistance for the livestream. Videos from the meeting are posted on the FSH Society's YouTube channel.

NEXT FSH SOCIETY PATIENT SUPPORT GROUP

Saturday, July 27th, 12:00-3:00 PM.
Kennedy Krieger Institute Outpatient Center
801 North Broadway, Baltimore, MD
Videos from the meeting are posted on the FSH Society's YouTube channel.

SESSION 1 (to be confirmed): Dr. Leigh Ann Curl of Johns Hopkins Hospital (team surgeon for Super Bowl champion Baltimore Ravens) will answer your questions about scapular fixation surgery.

SESSION 2: Panel discussion for young people living with FSHD. If you are in your teens and twenties, this meeting is for you. Topics will include adaptive sports, higher education and transitioning to living independently.

BREAKOUT SESSIONS: One session will be for young people to discuss topics of concern, whether it be dating and relationships or trying to persuade your parents to stop circling overhead... A parallel session will be held for the parents.

If you are interested in attending, please contact Lilleen Walters at LCWalters1207@aol.com or 703-835-1507. If you need local transportation, Lilleen will do her best to accommodate you. The on-site contact at the Kennedy Krieger Institute is Dr. Genila Bibat at 443-923-2697.

The meeting will be live-streamed for those who cannot attend. Details will be e-mailed closer to the date.

Pre-meeting opportunity: Patients and family members participating in research studies at KKI will be able to provide data during the hour preceding the meeting. Please contact Geni Bibat at bibat@kennedykrieger.org if you are interested in learning more about becoming a research volunteer.

April 25th, 2013, was a magical night! Besides being beautiful and balmy, this spring evening saw history being made at the Vancouver Art Gallery: BANKING ON A CURE was the first ever FSHD fundraising event to be held in our city! And given the huge success it was, it won't be the last!

The event was conceived by Madison Hooge, step-sister to FSHer Kristina McMullin. Both young ladies are 21 years old and have been part of each other's lives since 2007, a full year before Kristina was diagnosed with FSHD. Always impressed by Kristina's determination and focus, Madison felt her compassion for her sister grow and solidify as she watched Kristina process and accept her diagnosis, and continue to live her life and follow her dreams with unwavering strength, humility, grace and resolve. And a wicked sense of humor!

Madison's petite frame belies the size of her heart. Her enthusiasm is infectious and endearing. Her talent for event planning and mobilizing the people she needs is exceptional for someone so young.

She simply decided one day in February that she wanted to do SOMETHING to help bring knowledge about FSHD to the forefront. She wanted to be a part of what helps drive research forward, and she wanted to do it NOW. She wants to be a part of discoveries for treatments and a cure in time to have a positive impact on Kristina's life, and on so many others like her. And that's all it took: great love for another person, compassion for their hardships, determination, grit and a can-do attitude!


Here's what we accomplished:

- 250 guests paid to come to the event. We offered tiered ticket pricing to make it attractive to the younger people
- 90 auction items were procured and fetched over \$10,000 in revenue!
- 2 fabulous raffle items: an iPad 4 and a private plane trip raised close to \$10,000!
- 4 Black Jack tables (with professional dealers and play-money) were offered as entertainment.
- No-host bar: \$5 cosmopolitan cocktails, beer and wine. Complimentary water.
- Fabulous food: passed appetizers, stationary trays, and delicious desserts.
- A Live DJ kept a good vibe going throughout the evening!

Madison delivered a passionate speech about her journey to this event. Kristina spoke candidly and inspirationally about her life with FSHD. Dr. Daniel Miller from the University of Washington in Seattle gave a fabulous, audience-appropriate talk about the disease and the joys and sorrows he experiences with his research and clinical work.

We have received excellent feedback on the uniqueness of the concept. People really like the 'all ages' aspect.

Many thanks to the local Vancouver FSH-ers (Darren Church, Asifa Lalji, Ann Hardon, Keith Martin) and their families who stepped up to support the event. Although final accounting has not yet been completed, we estimate to have made approximately \$36,000!

We hope this event can start some momentum for others in Canada. If we all do our part, and work together, we CAN make a difference!! 

Editor's note: *Banking on a Cure* raised funds for the FSHD Canada Foundation, which works with the FSH Society to direct grants to high-quality scientific research.



Sisters in arms: Madison Hooge and Kristina McMullin



Stanford University Medical Center Hospital launches neuromuscular clinic with focus on FSHD

by JENNIFER FOWLER, COMMUNITY OUTREACH LIAISON, STANFORD HOSPITAL NEUROMUSCULAR CLINIC
Stanford, California

Our new multidisciplinary neuromuscular clinic at the Stanford Hospital re-launched in the Spring of 2013. The FSH Society helped us reach out to patients in Northern California, and we were so pleased to see a number of members, including the Society's Chairman of the Board, Dr. William Lewis, Sr. at our grand opening!

The neuromuscular clinic is led by John W. Day, M.D., Ph.D. Dr. Day oversees both the adult and pediatric clinics and is actively involved in neuromuscular research projects at Stanford University Medical Center. Additionally, Dr. Day is actively building a consortium of researchers at other institutions, so our patients have the most up-to-date information about research and how to get involved with clinical trials taking place locally and beyond.

The clinic has its own dedicated space in the newly remodeled Hoover Pavilion. The adult clinic meets two days a week at the Hoover Pavilion, while the existing pediatric neuromuscular clinic at Lucile Packard Children's Hospital has been expanded and sees patients two days a week, allowing the team to see many more patients.

The focus of the multidisciplinary neuromuscular clinic is an integrated, team approach to patient-driven care. This team approach looks not only at the disease, but addresses the



Dr. John Day

individuals' needs over their lifespan, at home, work or school. This model of integrated care from childhood through adulthood is a valuable resource, and offers continuous support for both the patients and their loved ones.

At each visit, patients might see the physical therapist, occupational therapist, respiratory therapist, the licensed clinical social worker, speech and language pathologist, pulmonologist, case managers, clinical care coordinator, the neurologists, research assistant, the community liaison and other specialists as needed.

The new Hoover building is fully accessible for our patients. The parking lot is in front of the building, and offers improved access for disabled guests. Also, there is a pharmacy, a lab and a medical library for research on site. We are very excited about re-launching our growing clinic. Our goal is to make this multidisciplinary clinic the best patient-centered neuromuscular clinic in Northern California. Dr. Day is eager to accept new patients to the clinic. [FSHWatch](#)

For further information: Please visit the Neuromuscular Clinic's website at: <http://stanfordhospital.org/neuromusculardisorders>.

The Stanford Neuromuscular Program maintains a private Facebook page that people can join. Simply search Facebook for the page called "Stanford Neuromuscular Disorders."

Or contact: Jennifer Fisher, Neuromuscular Community Liaison, Hoover Pavilion, 211 Quarry Road, Suite 206, Mail Code 5992, Palo Alto, CA 94304. Phone: 650-497-5934. E-mail: jnfisher@stanford.edu

▶ Memphis rocks out for FSHD

Spicerfest is a music benefit whose purpose is to raise awareness of FSHD and raise money for research to find a cure. Jonelle Spicer has FSHD and wants to do all she can to help find a treatment. It is especially important because the disease also affects her grandmother, mother, and uncle.

Jonelle and her boyfriend Rudy have been raising funds for research on muscular dystrophy over the past four years. In 2012, they decided to start supporting the FSH Society and held the *First Annual Spicerfest*. On November 3, 2012, many friends joined in to support their cause at Murphy's pub in Memphis, Tennessee.

The event showcased some of the best music Memphis has to offer, including performances by Good Question, Mister Adams, Misti Rae, Marcela Pinilla, ObStRuCtioN Of POWER, Paving Funk, Eldorado and the Ruckus, The Po' Boys, The Rough Hearts, Whatever Dude, The Candy Company, and Special Shoes. Food was provided by Barley, Hops & Hogs. Many local vendors supplied silent auction items.

The first event raised \$2,450. Jonelle says "I feel so blessed to have such amazing people in my life who care to help us in our endeavor to help find a cure for FSHD." Spicerfest 2 will be held during the summer of 2013. Stay tuned.



OUR EBAY CHARITY AUCTION SITE

The FSH Society is registered (as the "FSH Muscular Dystrophy Society") on eBay's charity auction site. If you have an eBay seller's account, you can put items up for auction and direct from 10 to 100 percent of the proceeds to the Society. <http://givingworks.ebay.com/charity-auctions/charity/fsh-muscular-dystrophy-society/76296/>



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. Go to <http://www.razoo.com> and search for "FSH Society".

Join the FSH Society Legacy Circle*

by JUNE KINOSHITA, FSH SOCIETY, EXECUTIVE DIRECTOR
Lexington, Massachusetts




These are exciting times for those of us engaged in the fight against FSHD: recent scientific breakthroughs give us many reasons to be hopeful. We at the FSH Society are grateful for the ongoing financial support that has made these advances possible. If you have ever wished that you could do more to help, **I invite you to consider a planned gift to benefit the FSH Society.** There are many forms that a planned gift can take, from gifts of appreciated securities to trusts that can pay income to you or a loved one.

I would like to draw special attention to three options:

- **The Charitable IRA Rollover** allows individuals age 70 ½ or older to transfer up to \$100,000 from the IRA directly to qualified charities. By transferring part or all of your required minimum distribution (RMD) to the FSH Society, you will have a lower taxable income. (Important: Current legislation allows Rollover gifts for calendar year 2013 only.)
- **Bequest from Your Will or Living Trust** costs nothing now, yet is a flexible and easy way to make a gift larger than you may ever have imagined possible. You can designate a specific dollar amount, asset, or percentage of your estate. Often, a codicil to your existing documents is all that is needed.
- Did you know that qualified retirement accounts can be among the highest-taxed assets in an estate? For this reason, many donors choose to **make a Beneficiary Designation.** In addition to retirement plan assets, you can designate the FSH Society to receive a percentage of a life insurance policy, savings account, or certificate of deposit – simply by updating a form.

To honor their lifetime commitments and to encourage others to step forward in making their own plans, we welcome donors who provide for the Society through their estate as members of the *FSH Society Legacy Circle*. If you have already included the Society in your plans, please let us know by returning the confidential membership form (download at the link below). Doing so will allow us to thank and recognize you for your gift intention, and your example may inspire others.

Before making any changes to your plans, we urge you to consult your financial and legal advisors. If you have questions about how your planning can support the work of the FSH Society in the future, please contact June Kinoshita (june.kinoshita@fshsociety.org, 781-301-6060). 

Links

FSH Society Planned Giving membership form — <http://www.fshsociety.org/assets/pdf/FSHSocietyPlannedGivingMembershipForm.pdf>

Charitable Giving Opportunities — <http://www.fshsociety.org/assets/pdf/FSHSocietyPlannedGivingCharitableGivingOpportunities.pdf>

*Formerly called the “Future Fund”.

2013 Events: Save the Date!

For details and tickets, visit <http://www.fshsociety.org/pages/conEvents.html>

July 5

Lakeside Celebration and Fundraiser for the FSH Society

Live band, dinner, wine and auction under a tent on the shores of gorgeous Gull Lake.
The Mackay Residence
5:00 p.m. – 10:00 p.m.
Hickory Corners, Michigan

September 7

Fourth Annual Fulmer Family FSH Society Benefit Dinner. Hawaiian theme and food!

Sharon Baptist Church
5:00 p.m. – 8:00 p.m.
536 North Ola Road
McDonough, Georgia

September 23

Swing for the Cure Golf Tournament (Benefiting FSHD Canada/FSH Society)

Pipers Heath Golf Course
Milton, Ontario, Canada

September 30

A Festive Evening of Music and Song

New York Botanical Garden
6:00 p.m. – 9:30 p.m.
Bronx, New York

October 4

Golf Tournament for FSH Muscular Dystrophy

Abilene Country Club
Abilene, Texas

October 5-6

Fourth Annual Celebrity Charity Walk ‘n’ Roll for FSH Muscular Dystrophy

Preceded on October 5th by a Fireside Chat with Dr. Gregory Block
Heritage Park
Irvine, California

October 18

Fifth Annual Cape Cod Walk ‘n’ Roll for FSH Muscular Dystrophy

Harwich Community Center
Harwich, Massachusetts



Genetic structure implicated in aging may influence *DUX4* expression

by DOUG CRAIG, Ph.D.
Emerson, New Jersey

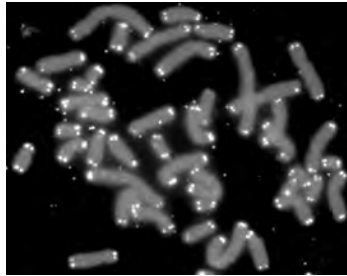
The telomere has been called the genome's "fountain of youth," a stretch of repetitive DNA located at the tips of every chromosome. This structure helps protect the integrity of genetic information. Without it, the genes can get miscopied, or fuse with other genes -- mistakes that can lead to cancer and premature cell death. But each time a cell divides and copies its DNA, the telomere gets shorter. The cell has ways to repair the telomere, but if it gets too short, the cell stops dividing. This seems to be one of the things that happen when we age, and scientists wonder whether keeping telomeres intact may contribute to a longer life.

Telomeres may play a key role in aging and cancer, and now, according to a new study, possibly in FSHD as well. Based on analyzing muscle cells from affected and unaffected members from two families, the study's authors report that the expression level of *DUX4* – the "FSHD gene" – appears to be influenced by the "telomere position effect" (TPE). The closer the gene is to the telomere, and/or the longer the telomere, the less *DUX4* gets expressed.

"FSHD may be the first known human disease in which TPE contributes to age-related phenotype," the study announced. Or in plain English, this discovery may help to explain why there is such a wide range in the age at which FSHD patients develop symptoms, and in the severity of those symptoms. If this finding holds up, it may have future implications for genetic counseling.

Although symptoms of FSHD generally first appear during the teen years in men and in the 20s for women, for some the first signs will not appear until age 40 or later. On the other hand, some have symptoms at birth. Similarly, while many patients will end up in a wheelchair, others may continue to walk, albeit with some difficulty, throughout their lives. Puzzling researchers even more, some individuals with the genetic markers for FSHD remain symptom-free throughout their lives.

DUX4 was recently identified as a gene that plays a key role in the development of FSHD. Multiple copies of the *DUX4* gene are located near the end of chromosome 4 – next door to the chromosome-4 telomere.



This discovery may help to explain why there is such a wide range in the age at which FSHD patients develop symptoms, and in the severity of those symptoms.

Because of its highly coiled DNA/protein structure, the telomere can entrap nearby genes – such as *DUX4* – preventing them from being used by cells to build proteins. One peculiar property of telomeres is that they become shorter with each successive round of cell division, and as the length decreases, so does their propensity to inhibit adjacent genes.

In the study "Telomere position effect regulates *DUX4* in human facioscapulohumeral muscular dystrophy," appearing in the May 2013 issue of *Nature Structural and Molecular Biology*, researchers showed that the expression of *DUX4* in muscle cells increased as the length of the telomere decreased. Furthermore, the effect of the telomere extended beyond *DUX4* to a neighboring gene, *FRG2*, which has also been implicated in FSHD.

These new findings indicate that telomere shortening, because of its link to the number of cell divisions, may be an important process for determining the timing of disease onset. Telomere length in human chromosomes is highly variable, and this could also translate into the individual variability in the onset and severity of the disease.

So far, this effect has only been confirmed using laboratory methods to adjust the telomere length in muscle cells from individuals with FSHD in two families and their unaffected siblings. Additional studies will be needed to show whether the effect of telomere length occurs in larger populations of individuals with FSHD and unaffected individuals with the FSHD genetic signature.

"We hope there will be prognostic value in being able to determine the specific telomere length near the contracted D4Z4

repeats [which contain the *DUX4* gene]," said lead author Woodring Wright, PhD. "This is technically a bit challenging, but hopefully we'll be able to do it within a year or so. If TPE turns out to relate directly to the age of onset and severity of symptoms, then it would have applications to genetic counseling and for identifying appropriate patient populations for clinical trials."

The current findings may also shed new light on FSHD. "We are hoping that other studies we are doing on how telomere shortening influences gene expression will help identify genes other than *DUX4* that are involved in the molecular pathogenesis of FSHD," Wright says. "Hopefully, knowing more about the genes involved will lead to better therapeutic interventions."

The study was conducted by an international team of researchers comprising the NIH NICHD Senator Paul D. Wellstone Muscular Dystrophy Cooperative Research Center for FSHD. The multi-disciplinary team of researchers involved in the study are from the University of Texas Southwestern Medical Center in Dallas, University of Massachusetts Medical School in Worcester, Massachusetts, Boston Children's Hospital, the Boston Biomedical Research Institute, the Hugo W. Moser Research Institute at Kennedy Krieger Institute and Johns Hopkins School of Medicine in Baltimore, and the Center of Excellence in Genomic Medicine Research, King Abdulaziz University, Jeddah, Saudi Arabia.

The study used genetically matched FSHD-patient samples available from the NIH NICHD Senator Paul D. Wellstone Muscular Dystrophy Cooperative Research Center for FSHD Research muscle biopsy collection at the University of Massachusetts

THE CENTERS FOR DISEASE CONTROL SPOTLIGHTS FSH MUSCULAR DYSTROPHY

... from page 1

ambulatory, suffering many falls and trips to the emergency room until he was reconciled to using a wheelchair. He regrets having put himself through so much to avoid the chair. "Now I can do so much more," he says.

To help public health officials understand the health care needs and quality of life of people with muscular dystrophy and their families, the CDC supports and manages MD STARnet, the Muscular Dystrophy Surveillance, Tracking, and Research Network. MD STARnet has identified and studied every individual born with Duchenne and Becker muscular dystrophy since 1981 in six states. The CDC is currently testing the feasibility of collecting information on the seven other muscular dystrophies including FSHD.

Working with the American Academy of Neurology (AAN), the CDC is also backing the development of clinical guidelines for four types of muscular dystrophy: FSHD, myotonic dystrophy, limb-girdle muscular dystrophy and congenital muscular dystrophy. These guidelines will be published by the AAN this year. The FSH Society will be joining other muscular dystrophy organizations to make sure doctors and patients everywhere know about the guidelines.

FSH Society President and CEO, Daniel Perez praises the CDC, saying "These are important initiatives to improve health outcomes and address health inequities for men, women and children with muscular dystrophy."

Over the 22 years since its founding, the FSH Society has worked closely with federal agencies, researchers, advocacy organizations around the world, and patients like Kevin Kirby, to raise and direct millions of dollars towards research on FSHD. That investment is beginning to pay off with the discovery of the genetic causes of the disease in 2010 and 2012. Pharmaceutical companies have teamed up with academic institutions to hunt for potential treatments.

Kirby turned 40 recently and in the same month became the owner of a van. "I don't think that's how your midlife crisis is supposed to go," he quipped. When asked if he thinks about the next ten years, Kirby says "I can't stress about the things I can't control. Today my life is great." **FSH Watch**

Medical School in Worcester, Massachusetts. The FSH Society works as the patient advocacy organization within and alongside the Wellstone Center and helps support patient education, recruitment, and travel when participating in the research study. Thanks to members' support, the FSH Society – NIH Wellstone cell lines now include 41 cohorts with 95 individuals, with two biopsy samples from each. The cell lines are available to researchers worldwide. **FSH Watch**

Reference

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<http://www.nature.com/nsmb/journal/vaop/ncurrent/full/nsmb.2571.html>

ERRATA WITH SINCERE APOLOGIES

The following donors were inadvertently left out of the 2012 Annual Donor Report. We deeply regret these omissions. We are grateful to all of the FSH Society's donors and thank you for support.

\$1,000-10,000

Benoit Allaire (was misspelled at Altaire)
Maureen, Rich and Liam Eye
Kelli O'Hara Naughton
Judith and Kenneth Seslowe

\$100-999

Ellen and Lawrence Allen
Mary Ellen Eye
Gerald and Marjorie Friedman
Andrew and Peggy Kahn
Wendy Levine
Joan Mark
Barbara and Robert Neustadter
Rhoda and Claus Pappenheimer
Carol and Erwin Riven (listed as
Jewish Communal Fund)
Jeffrey Rudikoff (misspelled as Rudikof)

Helpful Websites

CDC Features. Muscular Dystrophy: Kevin's Story — <http://www.cdc.gov/Features/MuscularDystrophy/>

CDC's muscular dystrophy website — <http://www.cdc.gov/ncbddd/muscular dystrophy/index.html>

MD STARnet — <http://www.cdc.gov/ncbddd/muscular dystrophy/research.html>

▶ Stella and Dot Trunk Show

My Stella and Dot trunk show on December 8th to raise awareness for FSHD was a lively and fun-filled event. People could drop by at anytime during the trunk show from 10:00 a.m. to 1:00 p.m. to purchase jewelry. The neat thing about Stella and Dot is that it is a sample sale so wares on display are not taken home by the buyers. That way people who come in towards the end of the show can still try on the jewelry. If a customer liked a particular piece of jewelry, they could order it and it would be shipped within five days. The trunk show was perfect to drop in and do some holiday shopping. The goal was to raise awareness of FSHD. When any person stopped by, I handed them the FSH Society flyers and contribution flyers. In addition, I had the FSH Society's Kelli O'Hara and Steven Blier promotional video playing in the background. We raised over \$700 in a short amount of time for FSHD research. The easygoing Caribou Coffee environment also helped to make people realize they were supporting a good cause. Next December I would love to aim for \$1,000. — *Carden Wyckoff*



Stella and Dot jewelry on display.

A LONG-AWAITED MOUSE OFFERS A NEW ANIMAL MODEL FOR FSH MUSCULAR DYSTROPHY

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serve an important purpose in drug development programs for FSHD,” remarked senior author Silvére van der Maarel of Leiden University in the Netherlands.

Other members of the team work at the Fred Hutchinson Cancer Research Center, Seattle, Washington, USA; King’s College London, United Kingdom; Radboud University Nijmegen Medical Centre, Nijmegen, Netherlands; and the University of Rochester Medical Center, Rochester, New York, USA.

The herculean project was initiated in 2003 by the FSH Society’s Marjorie Bronfman Fellowship grant. The patient-driven charity was seeking a definitive mouse model based on a genetic unit called D4Z4. Normally, people have ten or more of these units, repeated one after the other near the tip of chromosome 4. The majority of FSHD patients, in contrast, have fewer than ten.

The D4Z4 unit contains a gene called *DUX4*, which is toxic to muscle cells. In a series of landmark studies that unfolded over the past several years, an international team discovered that *DUX4* is normally only expressed during embryonic development and in the male germline (in stem cells that give rise to sperm). *DUX4* is not supposed to be active in other tissues, and the D4Z4 units act like bricks in a firewall, preventing the information in the *DUX4* gene from getting out.

In FSHD patients, however, the reduced number of D4Z4 weakens the firewall, allowing *DUX4* to be abnormally expressed in adult muscle in a quite remarkable pattern; only a small subset of muscle cell nuclei expresses abundant levels of *DUX4*. The result is devastating, as skeletal muscles degenerate, typically in the face (facio-), shoulder blades (scapula-) and upper arms (humeral), the anatomical areas initially affected that gave rise to the name of this dystrophy. Some patients endure hearing loss and/or abnormalities of blood vessels in the back of the eye.

The newly published mouse model contains 2.5 copies of the D4Z4 unit, a truncated number comparable to that seen in human FSHD patients. A second line of mice was created with 12.5 D4Z4 units, corresponding to an unaffected person. Both mouse models had high levels of *DUX4* in germline cells, as well as in embryonic stem cells and in developing embryos. Only in mice with 2.5 D4Z4 units, *DUX4* was also seen in all skeletal muscles, albeit at low levels and in highly variable ways.

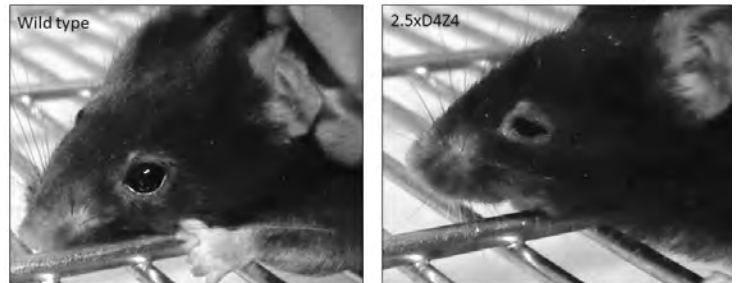
Further detailed analysis of *DUX4* expression revealed that mice with 2.5 D4Z4 units had the remarkable *DUX4* expression pattern seen in human FSHD patients: only a small subset of muscle cell nuclei expressed abundant levels of *DUX4*. In mice with 12.5 units of D4Z4, by contrast, *DUX4* was almost completely absent from muscles.

In terms of molecular activity, *DUX4* in muscle cells appeared to trigger other genes in networks similar to those previously reported in tissue collected from human FSHD patients. In addition, the D4Z4 units in these mice had reduced levels of “methylation”, which one can think of as the “mortar” that seals together the D4Z4 bricks in the firewall. With less methylation, the system cannot suppress the expression of toxic *DUX4*. The mice with 12.5 D4Z4 units had high levels of methylation, which was consistent with their low expression of *DUX4*.

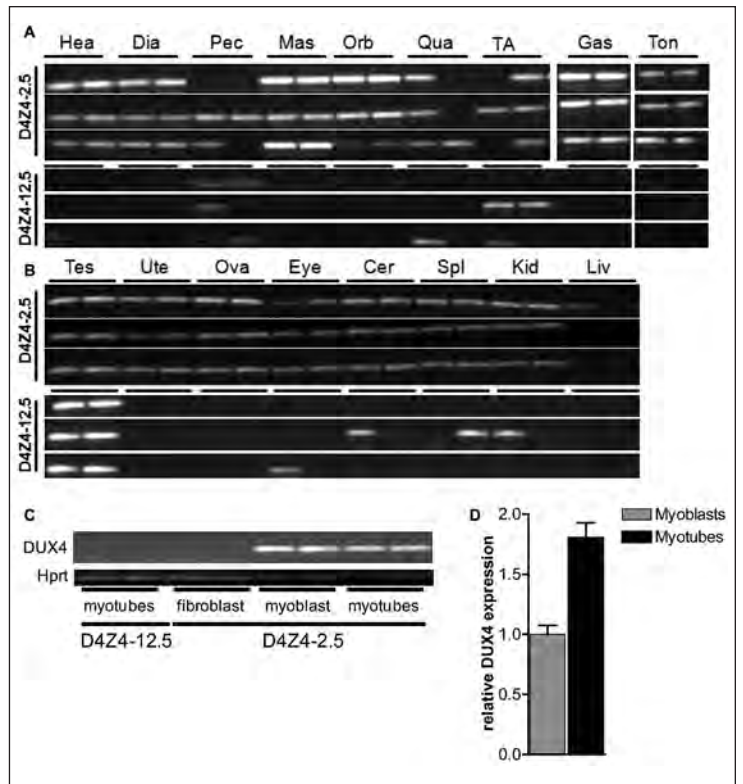
In other words, these “D4Z4-2.5” mice appear to faithfully mimic key features of FSHD. But the researchers warn that the differences between the 2.5-unit and 12.5-unit mice could

potentially be caused by as-yet-unknown differences in the genome sites where the D4Z4 units were inserted in the two mouse strains. In addition, it took the researchers 450 tries before they succeeded in generating the D4Z4-2.5 line, so there could be some unique mechanism at work in this line.

Nonetheless, the researchers are optimistic that this mouse will provide new insights into *DUX4*’s role in FSHD. For example, they hope to learn “why, how and when are sudden bursts of *DUX4* expression in skeletal muscle regulated.” In addition, they say, “As it was recently demonstrated that the detrimental effects of *DUX4*



A mouse with 2.5 copies of D4Z4, seen at right, developed keratitis (inflamed corneas). The wild-type mouse at left has normal eyes.



Analysis of transcriptional activity of *DUX4* in a panel of tissues of D4Z4-2.5 and D4Z4-12.5 mice.

DUX4 transcripts measured in 7 weeks old D4Z4-2.5 and D4Z4-12.5 mice (n = 3) in A) muscle tissue: Hea = Heart, Dia = Diaphragm, Pec = Pectoralis Mas = Masseter, Orb = Orbicularis oris, Qua = Quadriceps, TA = Tibialis anterior, Gas = Gastrocnemius, Ton = Tongue; and B) somatic non-muscle and germline tissue: Tes = Testis, Ute = Uterus, Ova = Ovary, Eye, Cer = Cerebellum, Spl = Spleen, Kid = Kidney, Liv = Liver C) *DUX4* transcripts measured in satellite-cell-derived myoblasts, myotubes and interstitial fibroblast extracted from EDL muscle of D4Z4-12.5 and D4Z4-2.5 transgenic mice. D) Quantitative RT-PCR data of *DUX4* expression in D4Z4-2.5 myoblasts (n = 2) and myotubes (n = 2) 48 hours after induction of differentiation. Errors indicate SEM of the plotted mean. doi:10.1371/journal.pgen.1003415.g003

expression in mouse muscle can be reversed by RNA interference, our model may also serve well therapeutic intervention studies targeting *DUX4* expression in skeletal muscle.”

The seed planted by the FSH Society generated additional major funding from other sources (including the Prinses Beatrix Spierfonds, National Institutes of Health, Muscular Dystrophy Association, Stichting FSHD, Friends of FSH Research, Fields Center for FSHD and Neuromuscular Research, the Gerald Norton Foundation and the Eklund Family) to complete the ten-

year analysis of the new mouse model. “I am very grateful to the Society for their longstanding support of our studies,” says Van der Maarel. FSH Watch

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Available online at: <http://www.plosgenetics.org/doi/1003415>



NEWS AND EVENTS

Friends Supporting Hope smashes fundraising record!!

Chris and Ellen Stenmon, their family, friends, and Society members gather to do good while having a good time

by **JUNE KINOSHITA, FSH SOCIETY, EXECUTIVE DIRECTOR**
Lexington, Massachusetts

When Chris Stenmon made it onto the Boston College High School wrestling team back in the late 1980s, he was eager to prove his athletic prowess at the sports-loving school. Yet hard as he trained, he wasn't getting stronger, and he eventually consulted an orthopedic surgeon at New England Medical Center.

“The surgeon took one look at me and left the room for five minutes and came back with a medical book with a picture of a teenager who looked very similar to me, with weak face, shoulder and upper arm muscles,” recalls Stenmon. It turned out that he had FSH muscular dystrophy.

The diagnosis came as a shock to Stenmon and his family. But he vowed to not let his disease pin him to the mat. In the 25 years since learning he had FSHD, Stenmon graduated from Boston College, became a C.P.A. and is now a Principal at the accounting firm O'Connor & Drew, P.C., in Braintree, Massachusetts. He also became active in the FSH Society, joining its Board of Directors in 2005.

On April 27th of this year, Stenmon rallied hundreds of supporters to the 15th annual “Friends Supporting Hope” fundraiser to benefit the FSH Society. Last year Stenmon raised a record \$38,000 and he was intent on surpassing that amount this year.

“Friends Supporting Hope” got its start fifteen years ago when Stenmon decided to celebrate the end of tax season – a grueling time in his profession – with a “pub crawl”



Chris Stenmon, Doug “VB” Goudie and Ellen Stenmon

to generate funds for the FSH Society. He sold t-shirts and raised \$1,000. Over time, the crowd snowballed. “One year I even met my future wife Ellen on the pub crawl,” he laughs. The couple now have two children.

A few years ago, the event morphed into a more traditional reception and auction. This year's event took place at Florian Hall in Dorchester, Massachusetts. Fox 25 News Commentator Doug “VB” Goudie presided as emcee. The Boston-based band Fenian Sons rocked the hall with its popular Irish sounds. As in past years, his wife Ellen took on the lioness's share of work, with help from a bevy of family members and friends.

Local companies lined up to sponsor the event, including South Shore Fuel and Fawcett Oil, Saint Anthony Shrine, Eastern Bank Foundation, Premier Mortgage, Rogers

Jewelry and Middlesex Bank. Lori Haverty, a spin instructor at the Reading Athletic Club, along with fellow instructors at Body Infusions in Wilmington, also got into the act, urging club members to join “spin-a-thons” to raise money for FSHD research.

On the night of April 27th, in addition to enjoying appetizers and drinks from a cash bar, guests bid on a variety of tantalizing silent and live auction items, including vacation packages, diamond jewelry, sports memorabilia and a helicopter tour.

With auctioneer VB Goudie pumping up the adrenaline, the live auction took an exciting turn when a bidding war erupted over a

unique, behind-the-scenes tour of Boston's famed transit system. The prize was offered by Stenmon's school friend, Richard Davey, currently the Secretary of Transportation for the state of Massachusetts. The tour includes an opportunity to pick the color for the lights illuminating the landmark Zakim Bridge. When the bidding topped out at \$1,050, Secretary Davey immediately offered a second tour to the competing bidder, netting the event an extra \$1,050. The audience burst into cheers and applause.

By the time the dust settled, this year's Friends Supporting Hope had raised more than \$43,000. For Stenmon, who turned 40 this year, beating his old record and reaching “Forty for Forty” was the best birthday present he could have wished for. FSH Watch

Product Review: The Zoom

by CALLE ERIKSSON
Stockholm, Sweden

My name is Calle Eriksson, age 40. I inherited FSHD from my father. My first memories of being like him were in fourth or fifth grade. I remember thinking I must have AIDS, as this was just around the time that AIDS was being talked about on radio and TV. I did not want to make the connection of having inherited FSHD.

Years have passed. I have had a really good and active life filled with various adventures, many of them involving snowboarding and traveling. As I knew my physical time was limited, I was in a hurry to explore the world. I was a professional snowboard photographer and magazine maker with the world as my office. I spent most of my adult life on the road traveling from resort to resort and had my base in the European Alps.

Finally FSHD and time caught up with me, and in 2007, my inner voice told me to slow down and move back home to Sweden. In 2009, I became a father to a wonderful little boy who is now four years old.

As my son's walking ability and agility increased, mine decreased. I could not see myself using a regular electric wheelchair with joystick, having to stay on paved roads and walkways. I wanted to be part of my child's adventures on his terms, exploring field and forest, snow and sand, hills and mountains. I needed something that would allow me to follow him, and catch up with him if need be.

I started my search for ways to keep up with him and came across a small ad in a magazine where they were "searching for pioneers for a revolutionary electric powered vehicle." The ad was talking to me and I applied.

In the very moment that I found the Zoom, my life changed for the better. My son cannot outrun me or out-bike me now. He has to climb a tree to get away! We can go to the beach on our own; the white sand is no problem. I just make a deal with other parents around to do minor rescues if need be.

In the winter, come snow, the permanent four-wheel drive lets me get around with no problems. Stockholm is covered with white five months of the year,



Calle Eriksson and his son on the [zoom]uphill

and lots of it. Other parents in a similar situation are stuck at home, but we are out playing and I take him to daycare all year 'round. Some mornings we face howling winds and 40 centimeters of snow. Days like this are now small adventures as they should be.

So how physically able does one need to be to use the machine? I'm 195 centimeters tall and use the wheelchair on a regular basis these days. I can still walk short distances under good conditions. It has been ten years since I could rise out of a chair or sofa without the use of my arms. I have the classic no biceps no triceps but a pretty okay grip. The Zoom requires very little arm power. The youngest buyer is four years old, and the oldest is over 70. It can be customized to fit kids, with a slower speed and less aggressive power.

The vehicle has a tremendous amount of torque and can climb steep hills gracefully. I use it every day as my number one transportation for all commuting. I use it in the subway system and other trains. It's nice with the big 16-inch wheels that can handle gaps between train and platform, and I always feel safe. I even go by bus sometimes.

The relatively low weight is also good when traveling: 70 kilograms with the

batteries mounted, it can be lifted by two strong people. The Zoom fits in the back of any regular station wagon. I have a Subaru Forester and can load the Zoom into it with two aluminum ramps. I also have a Volkswagon van, and this one I can drive straight into and jump off inside the car.

Service and maintenance

The chassis is what is unique about this machine. It is rock solid and made out of steel. If anything happens to it, it can be welded and repaired by any car garage. There are no high tech materials that require special expertise. The motors and electronics are standard e-bike parts that can be ordered from many dealers and even online.

My Zoom has been used daily now for two years, and the only problems I have had is a failed motor switch, which was replaced under warranty, and a handful of punctures and worn out tires. Not bad for having been used heavily with close to 6,000 kilometers on the odometer.

The past six months I have not even had a puncture after having filled the tubes with a product called Stans tire sealant. The tires are standard moto-x ones and can be found globally.

The notion of limitation is long gone, and my neurons are on fire looking for adventures in everyday life. Areas of my brain that had been shut down are yet again active and I feel like a twenty-first

century centaur, half man, half vehicle, with unlimited powers.

You can watch me ride my Zoom here: <http://www.zoomability.com/project/zoomare-calle-eriksson/> 

Editor's note: The FSH Society publishes product reviews as a service to FSHD patients. Views expressed are those of the reviewers. Publication of a review does not imply that the FSH Society endorses any products or services.

► Quick Facts about [zoom]uphill®

[zoom]uphill® has a permanent and patented symmetrical 4-wheel drive and is specially designed to be driven in rough terrain. The patented frame design allows all four wheels to keep contact with the ground no matter what type of underlying structure. By this means a continuous 4-wheel drive is maintained.

[zoom]uphill is an electric all-terrain vehicle. It can be driven at a maximum speed of 20 km/h (12 mph) and is possible to drive where you generally can walk without climbing. On sidewalks a “walking pace” of 5 km/h (3 mph) should be maintained if the laws in the specific country permits driving there. A bell and lights at front and rear are required when it’s dark outside.

[zoom]uphill is developed, designed and manufactured in Sweden with development and sales offices are located in Västerås.

If you are interested in experiencing zoom, get in touch with the company via its contact form or call +46(0)21-41 53 00. The price



for a [zoom]uphill is 7.700 Euro or 10.000 USD excluding VAT. Freight and packaging not included. Delivery time is 2 to 4 weeks depending on the volume of orders.

From the [zoom]ability website — full details at www.zoomability.com



NEWS AND EVENTS

Two-time Tony winner Judy Kaye to perform at Festive Evening of Song this year

Broadway supernova Judy Kaye will grace the stage at the FSH Society’s annual Festive Evening of Song. This year’s concert with two-time Tony Award-winner Kaye and legendary pianist Steven Blier will be on September 30th at the New York Botanical Gardens.

Kaye is an American singer and actress whose name has illuminated the marquees of Broadway in countless shows over the past four decades.

She has performed extensively in a staggering variety of roles including: Julie Jordan and Nettie Fowler in *Carousel*, Annie Oakley in *Annie Get Your Gun*, Nellie Forbush in *South Pacific*, Meg in *Brigadoon*, Hildy in *On the Town*, Lalume in *Kismet*, Lili Vanessi in *Kiss Me, Kate*, Pistache in *Can-Can*, Babe Williams in *The Pajama Game*, the Old Lady in *Candide*, Maria in *The Sound of Music*, Rose in *Gypsy*, Anna in *The Anastasia Game*, Aldonza in



Judy Kaye at the 2012 Tony Awards

Man of La Mancha, Lucy in *You’re a Good Man, Charlie Brown*, Sally in *Follies*, and Mary Magdalene in *Jesus Christ Superstar*.

In 1988, Kaye appeared on Broadway as Carlotta Giudicelli in *The Phantom of the Opera*, singing coloratura Ds and Es eight shows a week. She won the 1988 Tony Award, Best Featured Actress in a Musical, for this role.

In 2012, Kaye once again won a Tony Award for Best Featured Actress in a Musical for her role in *Nice Work if You Can Get It*.

With a three-octave range, Kaye “easily shifts between Broadway belt and soaring soprano” according to *Playbill*. Our September

30th Festive Evening of Song concert is not to be missed!

Ticket buyers and event sponsors, please contact fshconcert2013@fshsociety.org for further information. 



The FSH Society is ranked among “Ten Charities Worth Watching”

by JUNE KINOSHITA, FSH SOCIETY, EXECUTIVE DIRECTOR
Lexington, Massachusetts

The FSH Society, Inc., has just been ranked among America’s “Ten Charities Worth Watching” by one of the nation’s premier charity evaluators.

The FSH Society’s inclusion in the highly prestigious list was announced on Charity Navigator’s website. “Many of America’s most effective charities are also household names. But some well-known charities are less effective than you’d think, while a number of lesser known charities are truly exceptional. These 10 charities all operate on less than \$2 million a year, but they all earn a four-star rating from Charity Navigator,” the website states. “We encourage you to learn more about them.”

The FSH Society has received Charity Navigator’s four-star rating for five years in a row, placing it among the top four percent of U.S. charity organizations. The rating indicates that the Society “adheres to good governance and other best practices that minimize the chance of unethical activities and consistently executes its mission in a fiscally responsible way,” Ken Berger, President and CEO of Charity Navigator, wrote in a congratulatory letter addressed to FSH Society President and CEO Daniel Perez.

Based on information provided in IRS Form 990, Charity Navigator analyzed The FSH Society’s performance in seven financial metrics: program expenses, administrative expenses, fundraising expenses, fundraising efficiency, primary revenue growth, program expenses growth, and working capital ratio. It also evaluated the organization in several accountability and transparency performance metrics, giving the Society an overall rating of 68.5 points out of a maximum score of 70.

“Ninety percent of the members of our Board of Directors are personally affected by FSHD, so we care deeply about being accountable to the thousands of patients and families whom we serve,” said Perez. “We are delighted that we have been recognized for being highly efficient and effective. This exceptional designation from Charity Navigator demonstrates to the public that the Society is worthy of its trust.” FSHWatch

Links

Top Ten Charities Worth Watching — <http://www.charitynavigator.org/index.cfm?bay=topten.detail&listid=7>
The FSH Society on Charity Navigator — <http://www.charitynavigator.org/index.cfm?bay=search.summary&orgid=9927>



DOES THE SOCIETY HAVE YOUR CURRENT E-MAIL ADDRESS?

If you want to be sure to receive breaking news and other up-to-the-minute information from the Society, please send us your e-mail address at info@fshsociety.org.

GET SOCIAL!

Join our online communities to get news, ask questions, seek advice and support from fellow FSHD patients and family members. The FSH Society Yahoo! Groups forum, online since the 1990’s, 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 our homepage at www.fshsociety.org, click on the “Community & Reference” menu tab at the top of the page and select “Online Community” in the left hand vertical navigation menu. You’ll see links to take you directly to our Facebook page and Yahoo! Group. 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.

HAVE YOU MADE A GIFT TO THE SOCIETY IN 2013?

Thanks to the support from members like you, the FSH Society is a world leader in combating muscular dystrophy. It has provided more than \$4.4 million dollars in seed grants for pioneering research worldwide and has developed an international collaborative network of patients and researchers. Your generous support is making a real difference! Please return your gift in the enclosed envelope. Or contribute online at www.fshsociety.org. Thank you!

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.

4-STAR RATING

The FSH Society has been awarded its fifth consecutive 4 Stars by Charity Navigator and named one of America’s “Ten Charities Worth Watching”.

