

# FSH Watch



CONNECTING THE COMMUNITY OF PATIENTS, FAMILIES, CLINICIANS AND INVESTIGATORS



## CELEBRITIES JOIN OUR CAUSE

Meet the FSH Society's Honorary Board

by **JUNE KINOSHITA**  
FSH Society, Lexington, Massachusetts

Andy Warhol predicted that “In the future, everyone will be world-famous for 15 minutes.” In the era of reality television and viral videos, his words strike an eerily prescient chord. But it seems that FSHD is still waiting for its moment in the limelight. It's a disease that most of the public has never heard of.

So why not harness celebrity culture to help raise the visibility of FSHD? Working with retired Hollywood agent and nationally known event consultant David Mirisch, the FSH Society reached out to figures from television, film, sports, fashion, and music and secured 15 members for our newly formed Honorary Board.

These distinguished individuals include actors from beloved films and television

... continued on page 14



Max Adler

PHOTO: RICHARD WRIGHT



PHOTO: PAUL MUHLRAD

Jeff Miller and Peter Jones chat at the FSH Society's International Research Consortium meeting.



## International Research Consortium Meeting

Researchers gather on MIT campus

by **JUNE KINOSHITA**  
FSH Society

This past October 21-22, nearly a hundred researchers from around the world gathered at the David H. Koch Center for Integrative Cancer Research on the campus of the Massachusetts Institute of Technology for the annual FSH Society International Research Consortium meeting. Located at the world epicenter of biomedical research (forgive us for our Cambridge-centric perspective), there was a palpable feeling of FSHD research having “arrived” in the big time.

Conference host David Housman, PhD,

chair of the FSH Society's Scientific Advisory Board, began the meeting on a somber note by paying tribute to MIT police officer Sean Collier, who was slain in April only steps from the Koch Center by the Boston Marathon bombing suspects. Housman said, “Many of us knew him, and it was a very direct and personal experience. We are dedicating our publications this year to Sean.”

Around a dozen of the scientists and observers attending the conference were personally affected by FSHD. They included members of the FSH Society's “virtual journal club” of scientist-patients who have been meeting online to discuss current FSHD research.

Baziel van Engelen, MD PhD, of the Nijmegen Medical Centre in the Netherlands,

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## Looking Ahead

Hope and concern for what 2014 brings

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June Kinoshita

Dear Friends,  
 As we embark upon the  
 New Year, my heart is  
 filled with gratitude for the  
 remarkable year we have  
 just had.

In the past 12 months,  
 we have launched a series  
 of patient support group  
 meetings in Baltimore,  
 which is livestreamed over  
 the Internet to viewers everywhere. We have sent out  
 our first ever television public service announcements  
 to nine metropolitan markets across the U.S. Over  
 the summer, the Society recruited FSHD patients to  
 appear before a governmental committee reviewing  
 federal policies and funding for muscular dystrophy.

In this issue of FSH Watch, you can read about  
 some additional fruits of our labors. We have assembled  
 our Honorary Board of celebrities who will help raise  
 awareness of FSHD with the general public. On the  
 science front, FSH Society funding contributed to  
 the landmark discovery that the SMCHD1 gene is a  
 modifier of disease severity. At our annual international  
 scientific conference, the feeling of excitement about the  
 progress in FSHD research was palpable.

We are upping the ante on our support for research.  
 We reached out, and scientists responded with an  
 unprecedented number of exciting proposals. To keep  
 pace, the Society has also raised more money than ever  
 before, thanks to our dedicated volunteer event organiz-  
 ers, generous benefactors, and grassroots members.

We go into 2014 with great hope, but also with deep  
 concern for the challenges ahead. At the NIH, pay-  
 lines have dipped below the 10th percentile, meaning  
 that fewer than one in 10 grant applications receives  
 funding. The Muscular Dystrophy Association, as it  
 goes through its process of re-evaluating priorities and  
 directions within its strategic planning process, has as  
 of mid-December put a one-year hiatus on accepting  
 any new grant applications in basic and translational  
 research areas. This means that organizations like the  
 FSH Society are an essential lifeline to keep scientists  
 working in the FSHD field during these austere and  
 uncertain times.

None of these achievements would be possible  
 without our FSHD community, the most remarkable  
 and inspiring group I have ever had the privilege to  
 meet. I am profoundly thankful to them all: the Soci-  
 ety's staff, Board of Directors, and Scientific Advisory  
 Board, and the amazing patients and families who  
 volunteer their time to help advocate for FSHD,  
 organize fundraisers, write articles and letters, edit  
 videos, Tweet and Facebook our links ... the list is  
 endless, and so is our appreciation.

Thank you, and Happy New Year!

June Kinoshita  
 FSH Society Executive Director

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

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## 2013 FSH Society Pioneer Award

Honoring George W. Padberg, MD PhD

by **DANIEL PAUL PEREZ**  
FSH Society, Lexington, Massachusetts

The FSH Society is pleased to present the first Pioneer Award for outstanding contributions to research on FSHD to George W. Padberg, MD, PhD. We are profoundly grateful for his dedication to unlocking the mystery of FSHD and for his guidance, concern, and care for patients and their families worldwide.

The Pioneer Award was formally bestowed at Padberg's retirement lecture at Grote of St. Stevenskerk in Nijmegen, the Netherlands. There he has served as head of the Department of Neurology at St. Radboud University Medical Center.

Before George Padberg wrote his 1982 doctoral thesis on facioscapulohumeral disease, FSHD research was largely unattended to. Padberg's keen interest in the disease was due to first-hand experience with a patient. His thesis remains one of the most thorough and best references on FSHD.

Padberg established a gold standard in

FSHD investigation. With his comprehensive insights on the complicated nature of the disease, he helped many researchers and clinicians find a way to approach its genetics and clinical aspects, thereby opening it for further study and development.

In addition to pioneering ways to solve FSHD, Padberg is also one of the finest clinician researchers in neurology, skilled in his profession and craft, a man of vision, a consummate networker, and a significant force in bringing disparate groups together to change the status quo.

Padberg played a deeply significant role as a long-standing member of the FSH Society's Scientific Advisory Board, helping to pave the way for the Society to work with the international community and advocate for patient rights, research funding, and education worldwide.

For this, he truly deserves to be recognized as a Pioneer. All FSHD patients,



**George W. Padberg, MD PhD**

families, clinicians, and researchers owe him the deepest gratitude. [FSHWatch](#)



NEWS AND EVENTS

### TEERING OFF FOR A CURE

On September 23, 2013, we hosted our first annual Swing for the Cure golf tournament in honor of Jeff Johnston and in support of the FSHD Canada Foundation. Jeff was born and raised in Mississauga, Ontario, Canada, and now resides in Denver, Colorado. He has FSHD. His wife Beth is an active member and secretary of the FSH Society Board of Directors.

After attending the New York City benefit concert last year, which is co-chaired by Beth, we felt we could do our part to raise awareness and dollars for research in Jeff's hometown.

The event was held at Piper's Heath Golf Club in Milton, Ontario. The goal for the tournament was to have 72 golfers attend and raise \$25,000 for FSHD research. Thanks to the hard work and dedication from our volunteers, most notably Anna Johnston and Elaine St. Pierre, we exceeded our goals and had 94 golfers and raised over \$30,000.

Special thanks to all players, sponsors, FSHD Canada members, and volunteers who took time out of their schedules to make this day a success. A special thank you to the committee members who spent over seven months organizing the event: Jeff Johnston, Anna Johnston, Elaine St. Pierre, David Gunn, Paul Seymour, and Kevin Weisbrot.

We are looking forward to hosting the second annual Swing for the Cure tournament in September 2014. For more information visit our website at <http://fshdswingforthecure.webs.com/> or contact us at 416-550-0671.

— Dave and Steve Johnston



The winning foursome from Swing for the Cure. From left: Greg Purdy, Steve Johnston, Matt Larochelle, and Matthew Johnston.

# Genetic Modifier of FSHD Type 1 Found

Surprise! It's the gene for FSHD Type 2

by **JUNE KINOSHITA**  
FSH Society

**A**t last 2012 FSH Society's International Research Consortium meeting in San Francisco, scientists hinted that the then newly discovered genetic mutation for FSHD Type 2 may also make symptoms worse in patients with FSHD Type 1. (Over 95 percent of FSHD patients have Type 1; fewer than 5 percent have Type 2. The range and severity of symptoms are comparable for the two types.) That hint has now been confirmed and was published recently in the *American Journal of Human Genetics* by an international team.

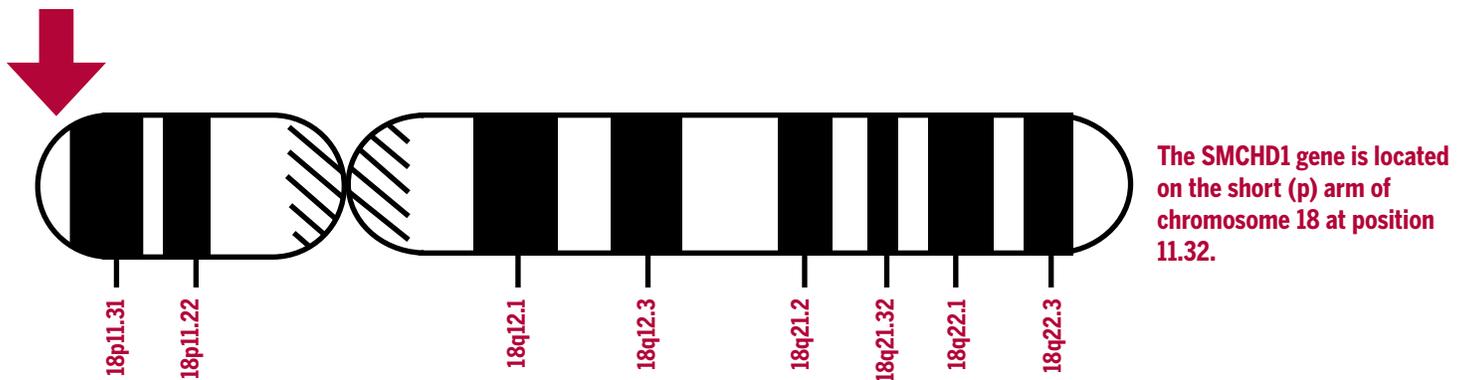
This finding bolsters what many researchers have long suspected: that there are “genetic modifiers” of FSHD—gene variants that make one person's symptoms more severe than those of a person who carries the identical FSHD genetic defect. This is a phenomenon familiar to many FSHD patients whose family members share

the barrier to loosen up and, just as in FSHD Type 1, allowing DUX4 to be expressed.

This is a satisfying discovery because it suggests that both types of FSHD are essentially identical, sharing the same molecular mechanism. This finding also strongly supports the hypothesis pointing at DUX4 misregulation as a key culprit and suggests a possible explanation for the wide variety of disease severity encountered in FSHD families.

Further studies are ongoing in France to establish the frequency of SMCHD1 mutations in a population of patients affected by FSHD Type 1. The outcome may help improve molecular diagnosis and genetic counseling for FSHD patients.

Moreover, “since DUX4 is involved in both diseases, all therapies that target DUX4 will be useful for FSHD Type 1, FSHD Type



the same FSHD genetic signature and yet have symptoms so mild as to go undiagnosed, while a sibling or child may be in a wheelchair.

FSHD Type 1 is linked to the loss of genetic material at the tip of chromosome 4. That area is made up of many units of a sequence called D4Z4. Normally, people have between 11 to over 100 D4Z4 repeats. But in FSHD Type 1, there are only between one and 10.

D4Z4 contains a gene called DUX4, which is normally silent in adult muscles. That's a good thing because DUX4 appears to be toxic to muscle cells. Scientists hypothesize that the repeated units of D4Z4 form a molecular barricade that prevents DUX4 from being “expressed.” But when there are not enough D4Z4 units, this barrier weakens and allows DUX4 to be expressed.

In FSHD Type 2, a different mechanism causes the disease. Type 2 patients have normal numbers of D4Z4 units. Instead, many have a mutation in a gene called SMCHD1 (pronounced “Smooch D1,” unless you want to intimidate your enemies and call it by its full name, “Structural Maintenance of Chromosomes flexible Hinge Domain containing 1”).

SMCHD1 is involved in attaching methyl groups to the D4Z4 units, shoring up the barrier to keep DUX4 from being expressed. But when SMCHD1 is mutated, its function is impaired, causing

2, and patients that carry both diseases,” said the study's lead author, Sabrina Sacconi, MD.

Sacconi was part of an international team with members from the University Hospital of Nice in France, Leiden University Medical Center in the Netherlands, the University of Padova in Italy, the University of Rochester Medical Center in New York, and the Fred Hutchinson Cancer Research Center in Seattle, Washington.

This work was made possible by many FSHD family volunteers and supported by grants from the U.S. National Institutes of Health, the Muscular Dystrophy Association, the Fields Center for FSHD Research, the Gerald Norton Foundation, the Eklund Family, the FSH Society, the Friends of FSH Research, the French Association Against Myopathies, the European Union Framework Programme, the Prinses Beatrix Spierfonds, and the Stichting FSHD. [FSHWatch](#)

## Reference

The FSHD2 Gene SMCHD1 Is a Modifier of Disease Severity in Families Affected by FSHD1. Sacconi S, Lemmers RJ, Balog J, van der Vliet PJ, Lahaut P, van Nieuwenhuizen MP, Straasheijm KR, Debipersad RD, Vos-Versteeg M, Salviati L, Casarin A, Pegoraro E, Tawil R, Bakker E, Tapscott SJ, Desnuelle C, van der Maarel SM. *Am J Hum Genet.* 2013 Sep 25. pii: S0002-9297(13)00379-0. doi: 10.1016/j.ajhg.2013.08.004.

# University of Massachusetts Medical School Launches FSHD Research Center

*New clinic to serve patients throughout Northeast*

by **JUNE KINOSHITA**  
FSH Society

**A** new, cooperative research center focusing on FSHD has been established at the University of Massachusetts Medical School (UMMS). The National Institutes of Health Senator Paul D. Wellstone Muscular Dystrophy Cooperative Research Center will focus its research and training programs on FSHD, becoming one of only a handful of such centers in the world.

The new center is the continuation of the Wellstone Center formerly headquartered at the Boston Biomedical Research Institute. The Center was recently awarded a renewal of the five-year, \$7 million grant from the NIH Eunice Shriver National Institute of Child Health and Human Development (NICHD).

Alongside the research center, UMMS has opened an FSHD clinic in partnership with Fairlawn Rehabilitation to serve patients from New England and across the northeastern United States. About 80 people attended an open house on November 16, 2013, at the gleaming new Albert Sherman Center in the heart of the UMMS campus. There they heard presentations about research, advocacy, and clinic services, and toured the laboratories. Patients and family members lined up eagerly to donate blood or saliva samples for a genetic research study.

There are currently no clinical trials of novel therapeutics for FSHD ongoing in the United States because of a lack of relevant preclinical data to direct development of therapeutic targets to disease mechanisms. The Wellstone Center at UMMS seeks to change that.

The Wellstone Center will collaborate with the FSH Society. The Society will consult on key communications, organize patient and research meetings and workshops, and provide education and support for patients and families participating in the Cell Core. The Cell Core is a tissue and cell repository of muscle stem cells and muscle biopsies, which the Center will make available worldwide to investigators to test new



**Home of the new UMMS FSHD Research Center**



**Patients and UMMS staff attended the open house at the Wellstone Center on November 16.**

FSHD therapeutics and to use the power of computational biology and genomics to establish an FSHD muscle biomarker database.

“FSHD leads to severe muscle weakness, and our Center’s therapeutic approaches will focus on development of drugs that target disease genes, as well as drugs that improve muscle strength,” says Center Director Charles P. Emerson Jr., PhD, professor of cell and developmental biology and neurology. “The therapeutic approaches being developed by our center will have broad application to the treatment of other muscular dystrophies and other debilitating medical conditions of muscle weakness including aging, muscle injury, and confined bed rest.”

“The new partnership between the Wellstone Center and other translational research groups at UMMS, such as the Department of Neurology and the Gene Therapy Center, has given the programs another opportunity to bring their unique talents to bear on a vexing clinical problem,” said Terence R. Flotte, MD, executive deputy chancellor, provost,

and dean of the school of medicine. “This is an important addition to the translational research enterprise.”

The UMMS Center’s mission includes training the next generation of muscular dystrophy clinical and basic researchers, and providing the broader research community with biomaterials for muscular dystrophy research. An important new initiative for the Center will be the establishment of the FSHD clinic, which will serve patients in the region and enable them to participate in research and clinical trials. The partnership among FSHD patients, their families, Center scientists, clinicians, and the overall industry will be essential for the Center to successfully carry out its mission to develop therapies for FSHD and other debilitating muscle diseases.

The Center also includes an Education and Training Core that will oversee a pre- and postdoctoral fellowship program designed to mentor young scientists for careers in muscle and muscular dystrophy research, with a strong focus on multidisciplinary training for FSHD and muscle disease research.

The UMMS Wellstone Center is one of six centers for muscular dystrophy research—and the only one solely dedicated to FSHD—funded by the National Institutes of Health, with additional funding from foundations and individuals. These centers engage in translational research with patients and industry to develop novel therapies for muscular dystrophies. A 2001 federal law, the MD CARE Act, directed the NIH to establish centers of excellence for research on muscular dystrophy; the program was subsequently named in honor of the late Senator Paul D. Wellstone of Minnesota, a champion of the NIH and muscular dystrophy research. [FSH Watch](#)

**The UMass Medical Center FSHD Clinic will see patients on Thursdays from 8:00 a.m. to 1:00 p.m. To make an appointment, call 508-334-2527.**

## September FSH Society Mid-Atlantic Patient Support Group

National experts talk about scapular fusion surgery and physical therapy

by JIM FOXX  
Bellingham, Washington



Leigh Ann Curl, MD

The Mid-Atlantic FSHD Patient Support Group had its fourth meeting at the Kennedy Krieger Institute (KKI) in Baltimore, Maryland, on September 28, 2013. I was able to attend, along with some 25 people, with another 20 watching live video online. Attendees included patients, family, friends, caregivers, and researchers.

In addition to seeing old and new friends, and sharing stories and concerns, the program included two exceptional professional speakers.

Leigh Ann Curl, MD, an orthopedic surgeon at MedStar Harbor Hospital in Baltimore, gave a detailed presentation on scapular fusion surgery. She has performed several hundred successful operations. Developed in the 1990s, it generally consists of tying and fusing the scapula to three or four ribs, restoring a firm foundation for full arm use.

The shoulder is an extremely complex joint with over 20 muscles controlling intricate and fine arm motion, Dr. Curl explained. Surgery is straightforward, but surgeons should be well trained and experienced.

Success depends on the surgeon's assessment of the candidate and the patient's ability to comply with a relatively long recovery process. It's not for everyone. Young candidates should have completed skeletal growth.

The surgeon must design the fixation based on each patient's unique situation. The four-hour operation has the patient fully anesthetized, face down. After opening the back (often through atrophied muscles), the scapula is wire-tied with bone-graft material (preferably from the patient's hip) to the appropriate ribs. The patient is commonly discharged from the hospital after three to five days.

The affected arm must not be used to carry any loads for six weeks, with minimal loads through 12 weeks, to give the bone grafts enough time to fully fuse the scapula to the rib cage.

With our weakened muscles, living without one arm may be extraordinarily difficult. Complications are rare, but could include infections, lung damage, and bleeding.

After one year, successful fixation leads to a dramatic improvement in qual-

ity of life, increased arm range of motion, strength, and reduced pain. Post-operative problems may include: broken fusion from premature loading, stiff shoulder, wire pain, or more rapid FSHD progression.

Our second speaker, Shree Pandya, PT DPT MS, flew in from the University of Rochester in New York. Pandya is a physical therapist on the neurological faculty. She is one of the most perceptive and FSHD-knowledgeable healthcare professionals I've met. She co-authored (with Wendy King) the FSH Society brochure *Exercise, Physical Therapy and FSHD*.

Pandya is active in promoting physical therapy specifically for FSHD patients, along with longer-term studies, because our progression extends over decades rather than months. Worth mentioning here is the National Registry for Myotonic and Facioscapulo-humeral Dystrophy at Rochester, a long-term collection of data from FSHD folks and their families using an annual status questionnaire. Now, after 12 years, we are beginning to see good results from "our" Registry. All of us should participate. Here's the link: <http://www.urmc.rochester.edu/neurology/national-registry/>

Shree Pandya noted that the goals of physical therapy are to maintain optimal health and wellness through adaptation or use, preventing or delaying secondary atrophy. Plans should be appropriate for each individual, for minimizing pain and fatigue while maintaining flexibility and strength. Orthotics, mobility devices, or assistive aids may be appropriate.

Benefits of physical activity are well known, but we need individual guidelines based on our specific situation. A physical therapist can be valuable, especially if he or she is willing to learn and work with us. We may need to provide specific guidelines, such as the FSH Society's brochure

Moderate aerobic exercise can be safe and improve our condition. The concern has been to do no harm and avoid the wrong exercise or too strenuous workouts that can lead to irreversible damage.

"Moderate exercise" is defined as "active but able to carry on a conversation comfortably," Pandya said. Two to five hours per week can be worked into our normal weekly routines and can include things like choosing to walk partway to a destination rather than riding in a car, or climbing stairs instead of taking the elevator.

Strength and flexibility training can improve our quality of life and delay FSHD progression. Simple exercises should be based on the grading of the current muscle strength on the widely used 1-5 scale (1 = No Strength; 3 = Overcome Gravity; 5 = Normal) and preferably start while muscles are still grade 4 or stronger.



Shree Pandya, PT DPT MS



**This patient cannot lift his arm to shoulder height on his left side because of the displacement of his scapula. Scapular fusion surgery on his right side restores the ability to raise his right arm.**

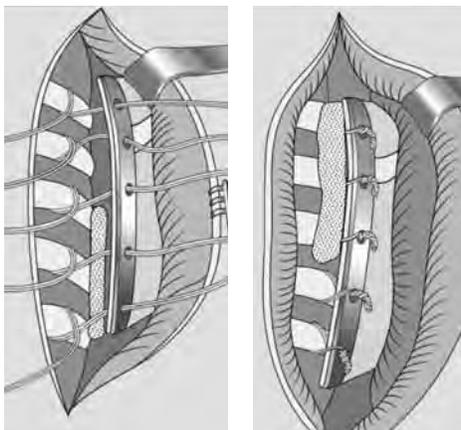
For strength training, stress is rated by the maximum number of times you can lift a weight. We should start with low weight, increasing to a weight that we can lift for a maximum of 10 times.

Strength exercises should be done daily. Many patients enjoy hydrotherapy exercises, which provide both buoyancy (neutralizing gravity) and gentle resistance. Elastic bands can be used at home. While we may not be able to lift our arms overhead, we can perform a full set of strength and flexibility exercises while lying in bed.

Pain can be an increasing problem. For example, standing for a long time can be excruciating. Using a barstool chair in the kitchen or a lower table at work can make a huge difference. Well-designed abdominal supports can help replace atrophied muscles.

Fatigue is another major issue. It can be reduced by conserving energy and working more efficiently. There is evidence that moderate aerobic exercise combined with cognitive behavioral therapy can also reduce fatigue.

Important factors for successful physical therapy include our current status (we vary as to age of onset and severity), our own



**In scapular fusion surgery wires are looped around the ribs, threaded through holes drilled in the scapular bone and a reinforcing plate (left). Bone graft material is sandwiched between the scapula and ribs, and the wires are tightened to fix the scapula into position (right).**

interests, and the facilities available to us. Implementation should include evaluation by a physical therapist who is willing to listen and learn about us as individuals, including specifics for FSHD. We can self-monitor, but we should give any program three months before re-evaluating with our physical therapist, Pandya advised. Start slow, give it time, exercise, and evaluate.

Ankle-foot orthotics (AFOs) are often our first assistive aids. There are many; some are better than others. Other devices include canes or walking poles, and many types of rollators. Mobility devices include a broad range, from lifts and scooters to power chairs. These (along with an adapted house and van) can involve considerable expense and life adjustments. Social embarrassment may be an issue.

Shree Pandya gave us many ideas to ponder. She is an example of how helpful a physical therapist, knowledgeable about FSHD (or willing to learn), can be to our own personal healthcare team.

At the end of the formal meeting, we socialized with researchers, patients, and new and old friends. [FSHWatch](#)

## GENETICS AND HEARING LOSS: Correlates with size of D4Z4 deletion

A recently published study from the University of Iowa reports that the likelihood of hearing loss in FSHD Type 1 (by far the most common type of FSHD) increases with the size of the D4Z4 deletion. Patients with between four and 10 D4Z4 units had no hearing loss. But among patients with between one and three D4Z4 units, one-third had high-frequency, progressive hearing loss in both ears, with onset in childhood.

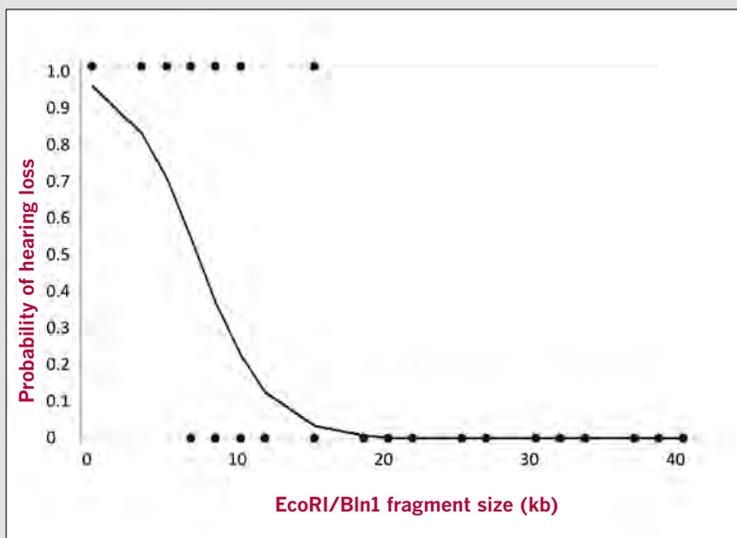
The authors of the study, which was published in *Neurology* in October 2013, conclude that the results support current recommendations for children with FSHD to be screened for hearing loss, even if the child passed a newborn hearing test.

“FSHD-associated hearing loss may occur after acquisition of language in preschool- and even school-age children,” write Basil T. Darras, MD, and Rabi Tawil, MD, in an accompanying editorial.

“Therefore, hearing screens should be continued until school age, particularly in young children with a small EcoRI/BlnI fragment size (less than 20 kb, i.e., one to three residual repeats).”

### Reference:

Lutz KL, Holte L, Kliethermes SA, Stephan C, Mathews KD. Clinical and genetic features of hearing loss in facioscapulohumeral muscular dystrophy. *Neurology*. 2013 Oct 15;81(16):1374-7. doi: 10.1212/WNL.0b013e3182a84140. Epub 2013 Sep 16.



# July FSH Society Mid-Atlantic Patient Support Group Meeting

## Transitioning to adulthood with FSHD

by **JIM FOXX**  
Bellingham, Washington

On Saturday, July 27, I attended the third Mid-Atlantic FSHD Support Group, which was held at the Kennedy Krieger Institute (KKI) in Baltimore, Maryland. This meeting is financially supported and sponsored by the FSH Society, thanks to donations from its members and benefactors. It was attended by some 20 FSHD folks, families, and friends, and over a dozen professionals working with FSHD. This meeting was focused on “Younger People with FSHD” around the theme of “Transitions to Independence.”

After an initial half-hour of socializing, the program began with Kathryn Wagner, MD PhD, giving an overview of the special situation teenagers with FSHD face. She spoke about how “coming of age” includes both social and medical transitions. At the KKI, medical support continues through the teen-to-adult transition. More typically, however, teens and young adults face disruptive transitions as they change doctors and schools, and move toward independence.

Dr. Wagner pointed the audience to some resources available to parents and individuals. (See list on page 9.) Although adapted from disorders other than FSHD, they are helpful. These include peer-support groups, adaptive sports, and mandated school, institutional, social, and workplace support requirements.

Every patient’s FSHD progresses at a different rate. However, when teens are struggling to be “normal” and fit in with their peer group, having a disability makes things doubly difficult. And increasing disability during the transition from family support to independent adult living likewise multiplies the challenges. Some colleges and universities can provide a supportive transitional environment.

Dr. Wagner cited a study by Schram et al. of Duchenne muscular dystrophy patients, which arrived at the following keys to a successful transition:

- Social and physical support
- Setting goals and reaching them (which promotes optimism)
- Acceptance, coping, and optimism
- Access to good medical and psychosocial information
- Continuity of medical care during the transition to adulthood

The transition must not be underestimated. It takes years and requires planning. Some cope through denial and are unable to deal with the overwhelming challenges all at once. Others can accept the fundamental realities, setting strategic goals and developing tactics to overcome specific challenges.

Many teens need to progress at their own pace, Dr. Wagner cautioned. Even adult mentors can backfire, overwhelming the teen who just isn’t ready. Adults and counselors need to assist with open communication but respect the teens’ pace, their need to know, and specific requirements for support. The good news is that most of us do get through it, finding our own adaptations, at our own pace. The goal (often hardest for caregiving parents) is independence.

Some specific suggestions for parents were offered:

- Facilitate open conversations on taboo issues (e.g., sexuality, separation from parental care, bodily appearance, loss of physical function)
- Teach skills for independence early

Dr. Wagner’s introduction was followed by a panel with two physical therapists, Carly Matichak, PT DPT, and Dorian Prince, PT DPT MSCS, and two local teens with FSHD, Colin Walters and Rebecca Abelman. They reviewed many specific social resources available, ranging from adapted sports to university or workplace access adaptations. (See resources below.)

For many of the sports activities, the underlying goals are

## Resources for Adapted Sports—Mid-Atlantic Region

Compiled by Carly Matichak, PT DPT, and Dorian Prince, PT DPT MSCS

**National Center on Health, Physical Activity, and Disability—Directories**

Website: [www.nchpad.org/directoriesIndex](http://www.nchpad.org/directoriesIndex)

### ► DISTRICT OF COLUMBIA

**MedStar National Rehabilitation Network**

Website: [www.nrhrehab.org/Patient+Care/Sports+Programs/default.aspx](http://www.nrhrehab.org/Patient+Care/Sports+Programs/default.aspx)

Handcycling (Tricia Nelson)

Email: [Patricia.e.nelson@medstar.net](mailto:Patricia.e.nelson@medstar.net)

Phone: 202-877-1427

Sled Hockey (Joan Joyce)

Email: [Joan.p.joyce@medstar.net](mailto:Joan.p.joyce@medstar.net)

Phone: 202-877-1420

WC Tennis (Brenda Gilmore)

Email: [brendagilmore@comcast.net](mailto:brendagilmore@comcast.net)

Phone: 240-460-5741

**The District of Columbia Department of Parks and Recreation**

Website: [www.dpr.dc.gov](http://www.dpr.dc.gov)

Phone: 202-698-1794

Activities: Adaptive sports and aquatics, arts and crafts, cultural events, and special events.

### ► MARYLAND

**Bennett Institute Physically Challenged Sports Program, Kennedy Krieger Institute, Baltimore, Maryland**

Email: [hermang@kennedykrieger.org](mailto:hermang@kennedykrieger.org)

Phone: 443-923-7844

Activities: Offsite programming may include participation in local, regional, national, and international athletic competitions. Special activity camps in golf, bicycling, and sled hockey are held on campus or at local university/recreational facilities.



building egos, confidence, and coping skills, as well as having social fun. With the right approach, those of us with disabilities can and do develop stronger than normal coping skills. Friends can be powerful support aides.

The benefits of sports rehabilitation are considerable. According to a survey by Disabled Sports USA, people who participate in their programs are more likely to be employed, be physically active, lead a healthy lifestyle, lead a fulfilling life, socialize more, and look forward to their future life. (See [www.disabledsportsusa.org](http://www.disabledsportsusa.org).)

At the end of our program, we welcomed a visitor, Elvina Sakellariou, PhD, a molecular biologist from the University of Maryland, who has been doing postdoctoral work growing human muscle tissue in mouse xenografts, with the hope of providing a new tool for understanding what goes awry in FSHD muscle. This is exciting, leading-edge work, funded in part by our FSH Society.

The meeting was livestreamed over the Internet (using UStream.tv) for offsite member access. Friends of FSH Research underwrote the advertisement-free livestream.

Video of the meeting is now available on the FSH Society YouTube channel and website. [FSH Watch](#)

#### Resources for transitioning to adulthood

As patients survive longer, many pediatric diseases are now confronting the issues of transition. Working groups' synopses and literature are available, some of which are also applicable to FSHD. Compiled by Kathryn Wagner, MD PhD:

**MDA Transitions Center**—[transitions.mda.org/](http://transitions.mda.org/)

**Parent Project Muscular Dystrophy**—[community.parentprojectmd.org/profiles/blogs/transition-discussion-lend-your-voice](http://community.parentprojectmd.org/profiles/blogs/transition-discussion-lend-your-voice)

**The Hospital for Sick Kids Good 2 Go Transition Program readiness checklists**—[www.sickkids.ca/Good2Go/What-we-do/Readiness-checklists/index.html](http://www.sickkids.ca/Good2Go/What-we-do/Readiness-checklists/index.html)

## INFORMATION AT YOUR FINGERTIPS

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

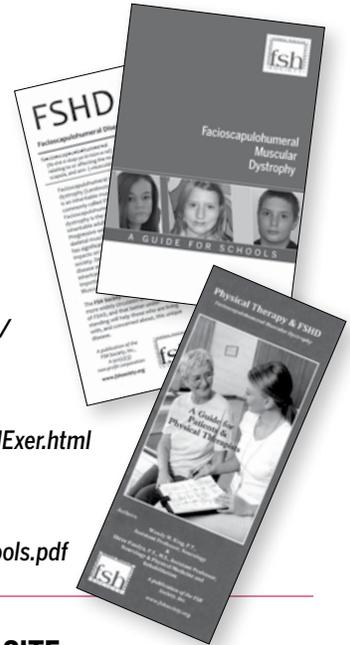
**FSH Society**  
450 Bedford Street  
Lexington, MA 02420  
Telephone: 781-301-6060  
Email: [info@fshsociety.org](mailto:info@fshsociety.org)

**FSHD Patient Brochure (in English)**  
[http://www.fshsociety.org/assets/pdf/PatientBrochure\\_May2009.pdf](http://www.fshsociety.org/assets/pdf/PatientBrochure_May2009.pdf)

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

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

**FSHD: A Guide for Schools**  
[http://www.fshsociety.org/assets/pdf/FSHSociety\\_FSHDPubs\\_AGuideForSchools.pdf](http://www.fshsociety.org/assets/pdf/FSHSociety_FSHDPubs_AGuideForSchools.pdf)



## 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/58335/>



#### Baltimore Adapted Recreation and Sports

Website: [www.barsinfo.org](http://www.barsinfo.org)

Email: [pam4bars@aol.com](mailto:pam4bars@aol.com)

Phone: 478-227-7386

Activities: Skiing, kayaking, camping, water skiing, skeet shooting, handcycling.

#### Chesapeake Region Accessible Boating (CRAB)

Website: [crabsailing.org](http://crabsailing.org)

Email: [info@crabsailing.org](mailto:info@crabsailing.org)

Phone: 410-626-0273

#### League for People with Disabilities, Hobbs Fitness Center

Phone: 410-323-0500

Activities: Fully accessible exercise and aquatics center.

#### ► PENNSYLVANIA

Pennsylvania Center for Adapted Sports

Email: [pcas@centeronline.com](mailto:pcas@centeronline.com)

Phone: 215-765-5118

Activities: Rowing (indoor/outdoor), swimming, sled hockey, skiing, handcycling, indoor climbing.

Pennsylvania Council on Therapeutic Horsemanship (PACTH)

Website: [www.pacth.org](http://www.pacth.org)

Email: [pacth@comcast.net](mailto:pacth@comcast.net)

Phone: 717-552-1628

#### ► VIRGINIA

Wish-A-Fish Foundation

Website: [www.wish-a-fish.org](http://www.wish-a-fish.org)

Activities: Multiple locations throughout Virginia, Maryland, Delaware, and Pennsylvania for fishing.

## 2013 Hustle for Muscle Golf Tournament

*Doubling up in year two!*

by **WENDY SHACK**  
Abilene, Texas

I cannot believe I am sitting here writing about the second annual Hustle for Muscle Golf Tournament. It seems like we were just finishing up last year's event. Time seems to go by so darn fast.

During last year's event, we learned that a new grandchild was on its way. This child would have no FSHD. Thank God for the research and work everyone did for this to happen. So we have a new Hustle for Muscle member on our team: Piper Kennedy Shack. We cannot express in words how this one little girl drives us every day to work harder for a cure. Talk about motivation!

This year we went to two tee times so that we could double the number of golfers. We hoped it would fill up, and fill up it did! We were even one team over for the last flight.

So many people were willing to give their time and money to come support the FSH Society. It was overwhelming and humbling. There was such a great spirit of giving and happiness for each golfer and volunteer who was there; it brought tears to our eyes. And what was more amazing was that 99 percent of our golfers from last time showed up again this year. All we can say about Texas people is when they are in, they are in!

Last year we had only a silent auction. This year we added a live auction to the evening event. The excitement generated by live bidding was amazing, and each auction item brought in more than we could have ever imagined. We will be doing this every year from now on.

One of the silent auction gifts was a handmade wooden Farley Boat. This boat was built by my husband Wayne and myself in Port Aransas, Texas. (See sidebar.)

We were blessed to have Doris Walsh from the FSH Society and her sweet husband Mike attend the event. Jim Chin Sr. was also able to attend to represent the Board of Directors of the FSH Society.

Through the years of our being involved, the FSH Society has become our family. Doris has become a wonderful friend. She told our family that we have been working together since 2009. Boy, how time does fly! These people were such a blessing to have. Everyone who met them was touched by them. They represent the FSH Society with great dignity and such giving hearts.

We also met a wonderful family that has been affected by Charcot-Marie-Tooth disease. These precious children, Kaleb and Addy, are called Kimbro's Angels. We cannot tell you how much these children touched the hearts of everyone who met them. Kaleb is a great spokesperson for Charcot-Marie-Tooth and is the future voice, I assure you, for his disease. You can find Kimbro's Angels on Facebook to learn more about this amazing family. As our family told them, we are all in this together!

Words cannot really express our family's appreciation to the FSH Society. The work they do is unbelievable. They have made our journey so much easier. We never feel like we are alone with this disease.

We are already putting our heads together for the Third Annual Hustle for Muscle Golf Tournament and looking for that great new thing to add to the tournament. We are looking forward to next year with such gusto. And, of course, we are always praying for a cure.

In closing, we just want to say with confidence and total faith that a cure is around the corner. The FSH Society works hard for all of us. Let's work hard for them! 



**Tournament organizers pose in front of the Farley boat they built for the auction.**

### The Love Boat

#### A CLASSIC VESSEL RAISES \$\$ FOR FSHD

The Texas Skiff first appeared on coastal shores during World War I. For nearly 60 years, these boats were handmade by craftsmen for the local fishing trade. But then, quality wooden boats gave way to cheaper, fiberglass boats, and a part of Texas history was nearly lost.

For four decades, people talked about the old Farley boats that a father and his two sons built in Port Aransas, Texas. In 2011, nearly 100 years after the first Farley boat was built, the newly reconstituted Farley Boat Works opened its doors to bring back the art of building wooden boats.

These Texas Skiffs are nearly 16 feet long and four feet wide. Rather than being built with cypress (as was originally done), the new Farley boat is made from mahogany. The gunwale rails, seats, transom, stem, sides, and bottom are all mahogany. And it is handmade, just as they were in the early 1900s.

God opened a door in October 2012 for a Farley boat to be built for my husband and myself as an auction item for the Hustle for Muscle Golf Tournament. We knew from the beginning that this is what God wanted us to do. We felt this would raise much needed funds for the FSH Society for research.

In August 2013, our journey began. We were guided by Darrell Lynn, the Farley Shop boat builder, and helped by many generous volunteers. Many good things came out of this experience: creating a beautiful boat, making lifelong friends, and making money for a wonderful organization. A win-win for everyone!

This has been a wonderful experience for us. We had a lot to learn and developed a new respect and love for this little boat. Our hopes are that someone loves and cares for her just like we would. And that her new owner will know that she was built with love by everyone who was involved.

—Wendy Shack



**Wendy Shack poses proudly with 94-year-old boat builder Bubba Milina in front of the Farley boat built for the auction. "He was a great encouragement to me. Just love this guy!" she says.**

## 2013 FSHD INTERNATIONAL RESEARCH CONSORTIUM MEETING

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launched the first session by offering some good news. In a controlled clinical trial, a combination of aerobic exercise and cognitive behavioral therapy (CBT) resulted in a significant reduction in chronic fatigue. A majority of FSHD patients have reported that fatigue is very detrimental to their quality of life, so this intervention could benefit many patients. The study is currently under review for publication in a journal.

The exercise and cognitive behavioral therapy protocols used in this study were described in Voet et al., *BMC Neurology* 2010;10:56, available online at <http://www.biomedcentral.com/1471-2377/10/56>.

In addition to non-pharmacological interventions, there is interest in knowing whether drugs currently on the market could be helpful. Because inflammation is suspected of playing a role in FSHD, Sabrina Sacconi, MD, and her colleagues at University Hospital of Nice in France wondered whether there might be any historical medical data to show whether anti-inflammatory drugs would slow down FSHD.

The investigators examined the medical records of 10 FSHD Type 1 patients who had initially been misdiagnosed as having polymyositis, a muscle disorder that is typically treated with high-dose prednisone (a potent immunosuppressant) for at least six months. Unfortunately, these patients' Clinical Severity and Manual Muscle Test scores got significantly worse during treatment, as compared to the period of 24 to 48 months after treatment was stopped.

"In my opinion, we have to understand the role of inflammation in FSHD before starting a therapy," said Sacconi. "This therapy may not only be unable to prevent progression, but can also contribute to worsening the disease."

However, prednisone "is not the best drug to address some of the inflammatory issues involved with FSHD," observed Stephen Tapscott, MD PhD, an investigator at the University of Washington, Seattle. It seems we can anticipate further research on anti-inflammatory drugs for FSHD.

### Novel treatment strategies?

Tapscott, who served as conference co-chair, noted that one of the remarkable

aspects of this year's meeting was the "striking agreement on DUX4" as a critical player in FSHD.

As a result of this consensus, many research teams are now searching "upstream" of DUX4 for drug targets that regulate the expression of DUX4, as well



**“We have to understand the role of inflammation in FSHD before starting a therapy. This therapy may not only be unable to prevent progression, but can also contribute to worsening the disease.”**

SABRINA SACCONI, MD, UNIVERSITY HOSPITAL OF NICE

as "downstream" for toxic processes that are set in motion by DUX4. A number of intriguing ideas were presented. These include:

- **Sex hormones.** Alberto Rosa, MD PhD, and his colleagues in Cordoba and Buenos Aires, Argentina, reported that DUX4 appears to modulate the activity of the progesterone receptor in cell cultures. The researchers found that estrogen in particular protected cells against DUX4 toxicity. Progesterone and testosterone were also protective, but not to the same degree. This finding aligns with the observation that menopause seems to trigger a worsening of symptoms in women and may account for why females generally seem to have milder disease than males.
- **Poly(ADP-ribose) polymerase 1 (PARP1).** Yi-Wen Chen, DVM PhD, and her team at Children's National Medical Center in Washington, DC, identified PARP1 as a possible target for controlling DUX4 expression. An enzyme involved in several important cellular functions, PARP1 interacts with the regulatory region of the DUX4 gene and increases DUX4 expression in FSHD cells, but not in normal cells. Chen speculated that in normal cells, the genomic region around DUX4 has a structure that doesn't allow PARP1 to interact. The investigators used fisetin, a PARP1 inhibitor found in many foods such as strawberries, apples, and onions, in cell cultures and

found that it reduced DUX4 expression. What's more, a PARP1 inhibitor called 3-aminobenzamide suppressed a downstream target of DUX4, but "we have not yet run experiments to see if PARP1 inhibition actually spares cells from DUX4 toxicity," Chen said.

If PARP1 proves to be a compelling therapeutic target, there are a number of PARP1 inhibitors in clinical trials for cancer. (In the meantime, an apple a day keeps the doctor away!)

- **Bromodomain and extra terminal (BET) proteins.** Fran Sverdrup, PhD, of Saint Louis University described a study that screened drugs targeting "epigenetic modifier proteins" to look for any that affect DUX4 expression in an FSHD cell-based assay (in collaboration with Stephen Tapscott and colleagues). This approach identified the "bromodomain and extra terminal" (BET) family of proteins as a key target involved in DUX4 expression. Much work still needs to be done to nail down the role BET proteins play in regulating DUX4 expression. These results are intriguing because BET proteins are a hot area in cancer, inflammation, and cardiovascular disease research, and a number of BET inhibitors are currently being developed as new classes of anticancer and cardiovascular drugs.
- **Beta-catenin and the Wnt signaling pathway.** Rather than probing FSHD cells growing in test tubes, Christopher Banerji of University College London turned to computers to sift for clues in publicly available data from gene expression profiles of FSHD muscle biopsies. He homed in on gene networks corresponding to known biological

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# FSH Friends on Facebook

*A private place for friendship and caring*

by MIA ARCHULETA  
Granbury, Texas

It all began in 2011 on a cold February Colorado night when Stephanie Staley decided to organize her Facebook friends into “groups.” What she didn’t realize at the time was that she was not merely placing her friends into categories such as family, school friends, church friends, FSH friends, etc. She was actually creating a social forum for each of those groups.



It didn’t take Stephanie long to realize that her “happy accident” or, as she likes to joke, “blonde moment,” was taking on a life of its own. While all of the other groups faded away, FSH friends began multiplying.

And so it began—a group of about 15 were now members of the FSH Friends Facebook Group. Each of those friends began adding friends, and so on. Before long the group had blossomed into an international gathering place for those with FSHD. Some have even been able to meet in person.

Because of the sudden growth, Stephanie felt she needed someone to co-administer the group, and I gladly accepted. It was a chance for me to finally meet and get to know others who live in the world of FSHD.

Members began to feel that we finally had a home, a place to call our own. A place we could visit to laugh, to cry, to share, to vent. Anyone can Google FSH muscular dystrophy and find all sorts of scientific information, but there aren’t many private places for FSHers to feel normal and to discuss things that most people outside of our world don’t understand.

The format that Facebook offers for such groups was the perfect venue. We love to have fun. We love to joke. We love to hijack threads. We also love to support one another with practical life experiences.

It is more than a support group, however. FSH Friends is a community in which we share life together. We are a diverse group of people who come together with different experiences, outlooks, and philosophies, but we all have the common bond of FSHD.

As the group has grown (372 to date), some members have chosen to add family and support team members so that they can see that FSHD is not “just in our heads.” Some choose not to include anyone close to them so that they feel free to vent and discuss things openly without fear that their loved ones would be hurt by some of the comments.

This is why the group is a secret group, meaning that all content is hidden from the public, and the only way to join is to be added by a member or to send a private message to either Stephanie Staley or myself, Mia Archuleta. It is a safe place to speak freely.

Social media has opened up doors for us to see that, though we are unique, we are not alone, and we all can learn from our fellow FSHers. It has also allowed us to network and become



aware of the many fundraisers throughout the country that support the FSH Society and their quest for a cure.

If you have FSHD and would like to join the FSH Friends Facebook Group, please send a Facebook message to Stephanie Staley (<https://www.facebook.com/stefstaley>) or Mia Archuleta (<https://www.facebook.com/mia.archuleta>) with a request to be invited into the group. **FSHWatch**

## New!

### FACEBOOK GROUP FOR TEENS AND YOUNG ADULTS

Younger people who are coping with a new diagnosis of FSHD or the challenges of trying to live their lives to the fullest have needs that may best be understood by their peers. At the FSH Society’s suggestion, Carden Wyckoff has created a secret Facebook group for younger people living with FSHD. To join, please message Carden on Facebook (<https://www.facebook.com/cardendanielle6>) or email [june.kinoshita@fshsociety.org](mailto:june.kinoshita@fshsociety.org).



interactions and threw out any that are associated with ancillary processes such as muscle wasting, aging, atrophy, and inflammation. This computational winnowing produced a set of biological networks that Banerji thinks are uniquely associated with the disease process in FSHD. One of these centers on a gene called beta-catenin, which is part of the “Wnt signaling pathway” already under investigation for a role in FSHD. Fate Therapeutics, a San Diego-based biotech, is developing a Wnt7a analog that targets this pathway.

In addition to seeking upstream and downstream targets, a number of research teams are going after the DUX4 gene itself. Sunny Das from the laboratory of Brian Chadwick, PhD, at Florida State University described the use of TALENs (transcription activator-like effector nucleases), a technology developed recently that is generating great excitement because it allows DNA-cutting enzymes to be custom-designed to splice out a specific part of the genome. The Florida group is developing a TALEN to remove the D4Z4 array and another to disrupt the “permissive” polyadenylation

cancerous, or they may not grow in sufficient numbers to produce a functioning muscle. The cells may be required in such huge numbers as to be impractical.

Still, it is exciting to see so many technologies that once seemed utterly futuristic—stem cells, genetically reprogramming cells, genomic engineering, cell transplantation—reach a stage where it no longer seems like a pipedream to bring them together to engineer a treatment.

### Genetic and epigenetic mysteries

While the vigorous pursuit of therapeutic targets is understandably exciting, many other presentations served to underscore how much basic research remains critically important. “That DUX4 is a necessity, that’s agreed upon. That it’s sufficient is arguable,” cautioned David Glass, MD, Global Head for Muscle Diseases at the Novartis Institutes for BioMedical Research in Cambridge, Massachusetts. (Glass is a member of the FSH Society’s Board of Directors and Scientific Advisory Board.)

Indeed, a large-population genetics study published by Rossella Tupler, MD PhD, and her colleagues in 2012 reported that 1.3 percent of the general population carries the FSHD Type 1 genetic signature, even though

functioning proteins.

In a landmark study published earlier this year (see story on page 4), an international team reported that SMCHD1, the gene implicated in FSHD Type 2, also modifies disease severity in FSHD Type 1. The investigators are now trying to understand the mechanisms by which SMCHD1 mutations lead to FSHD.

Presenting at the IRC meeting, study co-author Richard Lemmers, PhD, of Leiden University Medical Center in the Netherlands said that SMCHD1 normally binds to the D4Z4 region, where it mediates “DNA methylation” (the attachment of methyl to the DNA molecule) in a way that suppresses the DUX4 gene. But in FSHD Type 2, the gene is mutated and doesn’t bind as strongly, reducing its ability to tamp down DUX4. So far, 52 mutations in SMCHD1 have been found in FSHD Type 2 families.

Using samples from the University of Rochester in New York and INSERM in Nice, France, the team found that reduced methylation (resulting from reduced SMCHD1 binding to the D4Z4 region) roughly but significantly correlates with disease severity. Frédérique Magdinier’s group from INSERM in Marseille, France, also found differences in DNA methylation

***If this “DUX4 knockout” strategy is successful, in principle one would be able to take FSHD patients’ cells, induce them to revert into stem cells, treat the cells with TALENs so they no longer express DUX4, and then transplant those cells back into patients where they would develop into disease-free muscle.***



signal that is needed for DUX4 to be expressed.

If this “DUX4 knockout” strategy is successful, in principle one would be able to take FSHD patients’ cells, induce them to revert into stem cells, treat the cells with TALENs so they no longer express DUX4, and then transplant those cells back into patients where they would develop into disease-free muscle.

This all sounds magical, and in reality there could be many bumps and pitfalls along the road. A TALEN might accidentally target other, needed parts of the genome. Stem cells might turn

the percentage of diagnosed FSHD patients is much smaller.

What’s more, it has been observed that within FSHD families, whose members can be assumed to share the same genetic mechanism, symptoms can vary widely in severity. Some family members show no symptoms at all and were found to be genetic carriers only after they volunteered in a genetic study to serve as normal controls.

The reason for this wide variability of FSHD symptoms, scientists suspect, lies in the elaborate genomic machinery that regulates how the genetic code in our DNA actually gets turned into

across the D4Z4 region between FSHD Type 1 and Type 2 patients and non-manifesting carriers.

With the loss of methylation in and around the D4Z4 region, the chromatin structure opens up, setting the stage for specific events that trigger DUX4 to become expressed. In myoblasts (immature muscle cells) from FSHD patients, this happens when the cells divide and multiply. Eugénie Anseau and her colleagues in Alexandra Belayew’s lab at the University of Mons-Hainaut in Belgium have identified some of the gene networks involved in this

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**CELEBRITIES JOIN OUR CAUSE**



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series, three Olympic gold medalists, a Basketball Hall of Famer, an international supermodel, Broadway luminaries, a multi-Grammy award winner, and, yes, even a reality show contest winner!

The first to join the Board were “Glee” actor Max Adler and Broadway star Kelli O’Hara. Both of them appear in our public service videos, which are now being aired across the country and are available on the FSH Society’s YouTube channel (just search YouTube.com for “FSH Society” to find our channel).

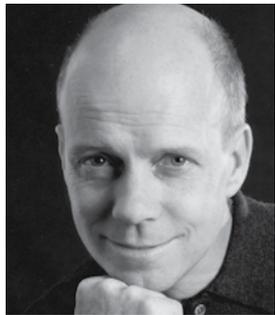
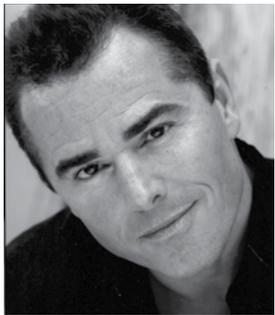
Please share these videos with your family and friends.



We look forward to working with all of our new Honorary Board members in the coming years to craft messages and campaigns that will touch the hearts and minds of the public. With their help, FSHD will become far better known—and not just for 15 minutes.

[FSH Watch](#)



				
<b>Max Adler</b>	<b>Kim Alexis</b>	<b>Sean Astin</b>	<b>Micky Dolenz</b>	<b>Brian Goodell</b>
				
<b>Scott Hamilton</b>	<b>Richard Karn</b>	<b>Christopher Knight</b>	<b>Florence LaRue</b>	<b>Pat McCormick &amp; David Mirisch</b>
				
<b>Rita Moreno</b>	<b>Kelli O'Hara</b>	<b>Stefanie Schaeffer</b>	<b>Mia St. John</b>	<b>Bill Walton</b>



MAX ADLER first drew wide attention for his role in the mega-hit FOX television series “Glee,” in which he played a closeted high school football player in one of the most dramatically acclaimed episodes of the series. He recently completed production on several feature films including “23 Blast,” a drama about a football player suddenly stricken with blindness; “Believe Me,” a comedy; and the family comedy drama “Saugatuck Cures,” in which he has the lead role. He has guest starred on other hit TV series including “CSI,” “Cold Case,” “Ghost Whisperer,” and “The Defenders.” Starting in 2014, he has a major recurring role on the award-winning ABC Family show “Switched at Birth.” Adler is a passionate activist for muscular dystrophy and appears in the FSH Society’s public service announcements. His mother and grandmother both passed away from the effects of FSHD.



KIM ALEXIS is among the most famous supermodels of all time. She has been featured on over 500 magazine covers since 1978. She has also appeared in numerous national television commercials and has been a spokesperson for many health and wellness companies. As a committed women’s health advocate, she has written five eBooks on health and wellness, available for the Amazon Kindle.

SEAN ASTIN starred in the Academy Award-winning feature film series “Lord of the Rings.” He is also known for his performances in “Goonies,” “Rudy,” “Encino Man,” “Toy Soldiers,” “White Water Summer,” and many other acclaimed films. Astin has appeared in numerous Emmy-nominated television series including “24,” “Jeremiah,” “Monk,” “Law & Order,” and “NCIS,” to name a few. He has also provided voices for well-known cartoon series including “Special Agent Oso” and “Teenage Mutant Ninja Turtles.”

MICKY DOLENZ was one of the stars and a lead singer for the iconic pop band and television show, “The Monkees.” He has appeared on numerous other television shows including “Peyton Place,” “Circus Boy,” “The Secret Files of the Spydogs,” and “The Tick.” In recent years, he has played Charlemagne at the Goodspeed Opera House for the revival of the musical “Pippin” and has appeared in the 2007 remake of “Halloween.”

BRIAN GOODELL won two gold medals for swimming in the 1976 Olympics at the age of 17, setting the world record in each event. He went on to win three gold medals for swimming in the 1979 Pan American Games. He was inducted into the International Swimming Hall of Fame in 1986, the Orange County Sports Hall of Fame in 1984, and the UCLA Hall of Fame in 1991.

SCOTT HAMILTON is one of the most acclaimed ice skating stars in the world. He is an Olympic gold medalist, TV skating commentator, actor, performer, producer, Emmy Award nominee, best-selling author, role model, humanitarian, philanthropist, and a cancer and brain tumor survivor.

RICHARD KARN is best known for his starring role of Al on the long-running television series “Home Improvement.” He has also appeared in major television series such as “CTRL,” “That 70’s Show,” and “Boy Meets World.” He was the host of popular game shows including “Bingo America” and “Family Feud,” one of the most famous game shows in television history.

CHRISTOPHER KNIGHT starred as Peter Brady on the classic television series “The Brady Bunch.” He also has appeared on “My Fair Brady,” “That 70’s Show,” “The Bradys,” “Another World,” “Joe’s World,” “Celebrity Circus,” “Entertainment Tonight,” “The Brady Bunch Variety Hour,” “The Surreal Life,” “The Brady Kids,” “CSI,” and many more. His feature film roles include “Fallen Angels,” “American Pie Presents: The Book of Love,” and “Letting Go.”



FLORENCE LARUE is an original member of the six-time Grammy Award-winning group The 5th Dimension, with number one hits such as “Up, Up and Away,” “The Age of Aquarius,” and “Wedding Bell Blues.” She starred in the national tour of Broadway’s Tony Award-winning “Ain’t Misbehavin’” and the theatrical production of “Mo’Magic.” She was named Woman of the Year by The City of Hope and also received the prestigious Spirit of Life Award.

PAT MCCORMICK is the only female diver in Olympic history to win four gold medals back to back in the 1952 and 1956 Summer Olympic Games. In 1956, she was the Sullivan Award Winner for “The Outstanding Amateur Athlete in the United States.” She holds 27 national titles and has been voted Athlete of the Year by the Associated Press, United Press International (UPI), and Sports Illustrated.

RITA MORENO has won four of the most prestigious awards in show business: an Oscar, a Tony, two Emmys, and a Grammy. Her roles span more than six decades, from her Broadway debut at age 13 to her role on the TV Land series, “Happily Divorced.” She has performed on Broadway, in London’s West End, appeared in more than 40 feature films, and has performed in regional theaters, most recently starring in her one-woman show, “Life Without Makeup.” She was awarded the Presidential Medal of Freedom in 2004 and the National Medal of Arts by President Obama in 2009. Her acclaimed memoir, “Rita Moreno: A Memoir,” was published in 2011.

KELLI O’HARA is one of Broadway’s beloved leading ladies. Most renowned for her stellar singing and acting, Kelli has starred in the Tony Award-winning revival of “South Pacific” at New York’s Lincoln Center. In 2003, Kelli earned her first Tony and Drama Desk Award nominations for “The Light in the Piazza.” She joined Harry Connick Jr. on Broadway in the 2006 Tony Award-winning production of “The Pajama Game.” Recently, she starred in the George Gershwin musical comedy “Nice Work If You Can Get It” opposite Matthew Broderick and received her fourth Tony nomination. She is starring in the Broadway production of “Bridges of Madison County.” In 2012, O’Hara performed with her friend and colleague, pianist Steven Blier, at the FSH Society’s New York benefit concert and appeared in the FSH Society’s public service announcement with Blier.

STEFANIE SCHAEFFER is a lawyer and leading businesswoman who rose to fame after winning the hit TV reality series “The Apprentice” in 2007. She was featured in Los Angeles Magazine and won a “Super Lawyers Young Rising Star Award” in both 2006 and 2007. Currently, she works for The Trump Organization and is in the process of launching a new video and book titled “Strut Your Stuff.”

MIA ST. JOHN is a WBC Boxing Champion and “the Sexiest Woman in Boxing.” She has appeared on all of the major talk shows. In 2008 she was presented with the WBC Goodwill Ambassador Award. She has also been acknowledged by the Mexican government for her role in sports and humanitarianism. Her latest book, “The Knockout Workout,” is in stores.

BILL WALTON, basketball legend, was elected to the Basketball Hall of Fame and named one of the 50 Greatest Players in NBA History. While at UCLA, he was a member of two NCAA championship teams, compiling an NCAA record 88 consecutive game-winning streak. He was a three-time All-American and College Player of the Year. He was also the winner of the Sullivan Award as top amateur athlete in the United States in 1973 and was the number one pick in the NBA draft in 1974. He was a member of the 1977 Portland Trailblazers and the 1986 Boston Celtics World Championship teams. In 1978, he was voted Most Valuable Player in the NBA.



# Champions Meet in Cambridge

*Second annual gathering generates ambitious goals*

by **KEES VAN DER GRAAF**

*President, FSHD Stichting and FSHD Europe, Wassenaar, the Netherlands*

Following on from the International Research Consortium and Planning meeting in Cambridge, Massachusetts, on October 21 and 22, the FSHD Champions met on October 23, 2013, to discuss the outcome of the research meeting and explore further ways for cooperation.

For me, the research meeting was an eye opener! More than 90 scientists from across the world attended the extremely well-organized meeting (thanks to Daniel Perez and June Kinoshita!). We listened to some 20 presentations from different research groups. On the second day we discussed the common ground. It was soon established that DUX4 plays an essential role in FSHD. This was a group conclusion, which in its own right was a breakthrough.

The Champions group, as explained in previous FSH Watch issues, consists of key representatives of all the FSHD funding agencies from around the world. Among them, the groups spent in 2012 US\$12.2 million on FSHD research. Organizations including the U.S. National Institutes of Health, L'Association Française contre les Myopathies (France), the Muscular Dystrophy Association (U.S.), and the Muscular Dystrophy Campaign (U.K.) attended alongside FSHD-specific organizations including the FSH Society, FSHD Global Research (Australia), the Chris Carrino Foundation for FSHD (U.S.), Friends of FSH Research (U.S.), the Shaw Family Foundation (U.S.), FSHD Europe, and FSHD Stichting (the Netherlands).

The Champions group is a network organization without a formal structure, bylaws, or official minutes, although it has a charter with goals. The meeting was convened by Daniel Paul Perez and myself. It is very inspiring that the group has produced in its first year of existence some quite useful reports, including an overview of all funded research projects, an inventory of research priorities at each funding agency, and critical success factors for clinical trials and mouse models. (I should say thanks to June Kinoshita, who has been very decisive in calling working group meetings and holding people accountable for the delivery of the tasks assigned to them.)



**The FSHD Champions met in the shadow of the MIT campus.**

This year's Champions meeting started with a presentation by Stephen Tapscott, MD PhD, from the Fred Hutchinson Cancer Institute in Seattle, Washington, who gave a summary of the most important conclusions from the International Research Consortium meeting.

David Glass, MD, executive director for muscle diseases at the Novartis Institutes for BioMedical Research in Cambridge, Massachusetts, and a member of the FSH Society Board of Directors and Scientific Advisory Board, gave a presentation on the importance of trial readiness. Given the fact that it is likely that we will enter into a phase where it will be necessary to test possible drugs or therapies on people with FSHD, we will need to ensure that the FSHD field has the knowledge necessary to design a successful trial. This includes understanding the natural history of FSHD—how the disease progresses in different individuals—and meaningful “clinical trial endpoints,” the gauge whose needle will tell us whether a drug is effective.

We agreed that this meeting of the Champions is a great vehicle for exchange, learning, and exploring ways for further cooperation. It was a no-brainer that we want to continue with the Champions meetings in 2014. June will act as the facilitator and coordinator. Daniel and I will continue as conveners of the meetings. There will be working groups that will deliver the agreed action points. They will meet by webinar

once a month.

On the action list there are projects such as the following:

- The development of a white paper on the key facts of FSHD. This will be published to encourage researchers to describe the disease in a uniform way.
- An overview of all the projects that were funded in 2013.
- An overview of the consensus view of the group on the most important research areas for the coming years.
- Sharing the most successful fundraising projects and ideas.
- A project to raise the awareness for FSHD globally, building on the best ideas from the individual FSHD funding and advocacy groups.
- A project to ensure that national registries of patients will be initiated in as many countries as possible, and are built on the same foundations and include a common minimum dataset.
- The development of a plan to address the issue of clinical trial readiness in a coordinated way.
- The development of a plan to promote FSHD to the scientific and medical communities to ensure we get ambitious young researchers to work on FSHD projects.

I personally felt very happy and inspired after the meeting. Real consensus was built, and the individual members showed an incredible willingness to start working on the different projects.

It is a very ambitious agenda, but that is what we need. We want to discover ways to halt the progress of the disease as soon as possible. We need to work together to achieve this. We agreed to do this, which is a crucial first step. We will have to work hard to deliver our promises.

The determination is there!

Thanks to June and Daniel for having organized this Champions meeting. 

**Editor's note:** *We thank Kees for his seminal work and significant contributions and impact on FSHD.*

“de-repression” of DUX4.

Daide Gabellini, PhD, of the San Raffaele Scientific Institute in Milan, Italy, discussed epigenetic modifications that regulate the expression of DBE-T (D4Z4 binding element transcript), a regulatory “switch” for DUX4 that is elevated in both FSHD Type 1 and Type 2. Gabellini’s team has reported that DBE-T binds to the FSHD site at the tip of chromosome 4, in combination with a third protein called ASH1L. He proposed that this event causes a structural change in the chromosome that permits genes in the region (DUX4,

Wright hypothesized.

While most research on FSHD genetics is focused on D4Z4/DUX4, the discovery of SMCHD1’s role in FSHD Type 2 could open up some fascinating new doors. Meena Upadhyaya, MD PhD, from Cardiff University in Wales, United Kingdom, identified mutations of SMCHD1 in a family affected by an unusual muscular dystrophy with some FSHD-like features.

“As SMCHD1 has a wider role in global genomic methylation, the possibility exists that it could be involved in other complex, undiagnosed muscle disorders,” she said.

“As SMCHD1 has a wider role in global genomic methylation, the possibility exists that it could be involved in other complex, undiagnosed muscle disorders.”

MEENA UPADHYAYA, MD, CARDIFF UNIVERSITY



among others) to be expressed. The team is currently working out the details of how DBE-T, ASH1L, and the D4Z4 region interact—a necessary step toward designing a method to block DUX4 expression.

Woodring Wright, MD PhD, of UT Southwestern Medical Center in Dallas, Texas, spoke about his novel hypothesis involving telomeres, special structures at the tips of chromosomes that are involved with maintaining the integrity of genetic information. Telomeres get shorter as organisms age and are thought to play a role in the aging process. Wright’s group had shown that DUX4 protein expression increased 10-fold in the nearby presence of shortened telomeres (see FSH Watch Spring 2013, p. 14).

Could shorter telomeres influence other genes that are farther away? It appears that they can, due to a mechanism called the “telomere looping effect.” Wright reported that in myoblast cultures, the researchers saw differences in gene interactions in FSHD patients’ myoblasts with shortened telomeres, as compared to those with long telomeres, which more resembled cells from unaffected, age-matched siblings. This difference “could explain much of the age-related onset and variability of the disease,”

This raises the intriguing possibility that genetic diagnosis could point to a broader family of diseases beyond FSHD that share underlying genetic origins.

### What makes DUX4 toxic?

DUX4 is a type of gene called a transcription factor, which means that when expressed, it causes other genes to get transcribed (the DNA code gets changed to an RNA message), setting off a multitude of biochemical cascades which, in the case of DUX4, are presumed to result in muscle damage.

But which of many such cascades is the cause of FSHD? That’s not yet known, but a number of potential culprits were discussed:

■ **Apoptosis.** Jacqueline Domire, Scott Harper, PhD, and their colleagues at Nationwide Children’s Hospital, Columbus, Ohio, presented their research pointing to a gene called p63, which is triggered by DUX4. P63 is involved in apoptosis, or “programmed cell death.” Apoptosis is a way in which biology eliminates unwanted cells—the webbing we once had between our fingers (in utero), or cells that are infected or pre-cancerous—but when

switched on at the wrong time and place, can cause healthy cells to die. Using cells from the FSHD collection at the NIH Wellstone Center at the University of Massachusetts, Harper’s team showed that p63 expression is significantly increased in FSHD cells compared to normal controls.

■ **Autophagy.** Cells rid themselves of garbage (misshapen and idle proteins, and worn out or broken parts) through a process called autophagy. There had been hints of reduced autophagy in FSHD, which could cause muscle cells to become diseased. Yi-Wen Chen, DVM PhD, of the Children’s National Medical Center in Washington, DC, presented data indicating reduced autophagy in biceps of FSHD patients, as compared with their less affected deltoid muscles.

■ **Germline and stem cell genes.** Previous studies have shown that DUX4 induces the expression in FSHD muscle of many genes that normally are only expressed in germline (sperm precursor) and stem cells. Fedik Rahimov, PhD, Louis M. Kunkel, PhD, and their colleagues at the Boston Children’s Hospital and NIH Wellstone Center at the University of Massachusetts reported that they analyzed a larger collection of tissue and cells from 52 FSHD patients and 40 unaffected family members, and found results consistent with the earlier findings. This gives researchers greater confidence that these genes are actually important in the disease process. These genes could serve as biomarkers and also point to potential targets for therapy.

■ **Ret enzyme.** Louise Moyle from the laboratory of Peter Zammit, PhD, at King’s College London reported that DUX4 activates the Ret co-receptor, which is normally involved in regulating the process by which myoblasts multiply and mature into muscle. Whether misregulation of Ret is a major factor in FSHD remains to be determined, but if abnormal Ret signaling turns out to play a role, blocking it could be a treatment strategy. Currently, drugs to inhibit Ret are being developed for cancer therapy.

... continued on page 19

# From X-Men to DUX4

Explaining FSHD without PowerPoint

by **GREG BLOCK**  
University of Washington, Seattle



**Gregory Block, PhD, at the Celebrity Walk 'n' Roll**

**T**his past October, I had the pleasure of addressing an audience of celebrities, donors, and patient advocates at the FSH Society Walk 'n' Roll held at the Dave & Buster's restaurant in Irvine, California.

The task was challenging: to educate people about the disease, some of whom had no background in genetics or had never heard of FSHD before. I was armed with only a microphone and a mole-skin notebook containing a madman's outline of talking points. I was wearing a black shirt, black pants, and a black tie, which also happened to be the standard Dave & Buster's uniform. After being accosted by customers with beer orders and complaints about broken game machines, I stood up on stage to deliver my message.

My message was simple. I wanted to tell people what FSHD is and why it is interesting and perplexing from a scientific perspective. Second, I wanted to convey that in order to study FSHD we've pushed our knowledge of biology and technology to the brink and continue to do so every day. Last, I offered my humble opinion that I think the disease is theoretically treatable.

I began by explaining that FSHD is a genetic disease, but unlike most genetic diseases, it is not caused by a single genetic mutation. Gene mutations are simple to understand; after all, they confer superheroes like the X-Men their superpowers. Recall the pinnacle villain from the comic/movie, Magneto, the mutant who is capable of controlling metal. Because metal is in everything, this sinister individual can stop flying bullets or rip the iron out of your blood. After he was caught by the X-Men, he was contained in a unique plastic prison embedded in a vast, empty warehouse.

I made the analogy that FSHD is the failure of a genetic plastic prison. The gene that causes FSHD, called DUX4, is present hundreds of times in the genome, within a motif called D4Z4. The multiple DUX4 genes are aligned in a head-to-tail orientation, like beads on a string, and that tells the genome to keep the gene silenced.

Keeping DUX4 silent requires the D4Z4 array to be long, usually over 10 repeats. Locking up DUX4 also requires a myriad of proteins, which we can think of as genetically encoded prison guards ... why not? When either the repeat structure is lost, or a "genetic prison guard" fails at his job, DUX4 escapes periodically and wreaks havoc on skeletal muscle.

Thinking this way about a disease is taking us into uncharted territories of biology, where we will learn even more, not only about FSHD, but perhaps other diseases as well.

After speaking with everyone in the room candidly about FSHD, I was left with a bit of an uneasy feeling. Although I've gone to school long enough to warrant being called "Dr. Block," I do not see patients, nor do I even have a permanent position at a university.

In the discipline of discovery, there are millions of questions to ask, even more ways to interpret the data, and more ways still to spin your discoveries into scientific papers. The end result of all this process is a lot of noise. The only way for a community to parse through the din is to work together.

At some point toward the end of the evening, I was asked whether I began studying FSHD because I had a personal connection to the disease. I thought about how many FSHD patients I've met over the last four years, and how many side hugs I've gotten. "No," I said. "But I do now." [FSH Watch](#)



## ADVOCACY

### Fighting for their future



**Angela Farkas nominated the FSH Society as a designated charity for her company's year-end gift. Her fellow employees at Adveq, an international private equity and real asset investments advisory service, selected the Society to receive \$10,000. Angela's daughters are both affected by FSHD. Thank you, Angela. We're working hard to reach the day when treatment can be offered to your beautiful girls.**

One of the big, unsolved mysteries of FSHD is the fact that the DUX4 gene message is expressed in no more than one in 1,000 muscle cell nuclei, but the protein it encodes is present in up to 40 times as many nuclei. (The DUX4 protein targets the cell nucleus, where it triggers the expression of downstream genes.)

Where is all that DUX4 protein coming from? In 2012, Alexandra Belayew and her colleagues at the University of Mons-Hainaut in Belgium proposed that DUX4 protein could diffuse from one nucleus to others. At the IRC meeting, Julie Dumonceaux, PhD, and her colleagues at INSERM in Paris, France, reported that this indeed appears to be the case.

The French team mixed human FSHD muscle cells with mouse muscle cells and detected DUX4 protein in mouse cell nuclei, where it shouldn't have been found because mice don't have the DUX4 gene. This meant that the protein must have traveled from the human cells into the mouse nuclei. What's more, the DUX4 protein in the mouse cells was able to activate some of the genes known to be targeted by DUX4 in humans. This "nuclear spreading" phenomenon may explain how the DUX4 gene can be so toxic, despite being expressed in so few nuclei.

### Muscles in a dish

From the University of Washington, Joel Chamberlain, PhD, described experiments using a gene shuttle to deliver a D4Z4 repeat from patient-derived DNA into mouse muscles. The muscle from these mice appeared similar to muscle from FSHD patient biopsies, with split muscle fibers and clusters of inflammatory cells surrounding single fibers. DUX4 mRNA and protein were expressed in the mouse cells, and further characterization of the model is in progress. Chamberlain said she hopes this mouse model will shed light on cellular pathways stimulated by DUX4 and prove useful for testing therapies.

Uli Schmidt, PhD, from Genex Biocells of Sydney, Australia, presented a new cellular model created from human embryonic stem cell lines. These stem cell lines came from donated pre-implantation genetic diagnosis (PGD) embryos carrying the FSHD deletion. The company developed a process to coax the stem cells to develop into skeletal muscle cells. "To our knowledge, this is

the world's first process that allows simple and robust industrial-scale cell production" of muscle, Schmidt said. The company is now comparing myotubes from normal and FSHD-affected cell lines.

### Gearing up for clinical trials

A major challenge for clinical researchers is predicting when a muscle is about to deteriorate in an FSHD patient. Having such a tool would be invaluable in a clinical trial, as researchers could track a vulnerable muscle to see if a treatment helps to prevent it from degenerating.

Enzo Ricci, MD, of the Institute of Neurology at the Catholic University in Rome, Italy, presented data showing that magnetic resonance imaging (MRI) is able to detect deterioration of upper-body muscles, notably the trapezius and serratus anterior. These can be seen through MRI to degenerate in an asymmetric pattern that is a hallmark of FSHD.

Intriguingly, some of the MRIs also revealed edema (increased water) in healthy-looking muscles that degenerated shortly afterwards, as documented by images made some months later. Ricci proposed that the edema indicates the presence of inflammation, which might be an initial stage in the degenerative process. Further studies are needed to determine whether edema can serve as an actual predictor of impending muscle loss.

Alongside imaging technologies, biomarkers that can be measured in blood

angiogenesis (proteins that stimulate the growth of blood vessels) show promise and will need a follow-up validation study to confirm their relationship to disease," the study's authors said.

In addition to biomarkers to detect disease and track the effects of therapy, FSHD clinical trials will require sensitive outcomes to measure the patients' muscle strength, ability to function, and quality of life. Rabi Tawil, MD, of the University of Rochester presented a poster with preliminary data showing that the six-minute walk test (which measures the distance a person can walk in six minutes) yielded consistent results and correlated well with composite scores on tests of leg muscle strength. This was true even in patients with minimal leg weakness. The preliminary data involved only 10 patients and will need to be confirmed in larger numbers of patients.

This is an incomplete sampling of the presentations that filled the first day of this year's International Research Consortium meeting. On the second day, the assembled scientists engaged in a lively discussion focusing on future priorities for FSHD research. We will be sharing the outcome of that discussion in the next issue of *FSH Watch*.

We thank the conference co-chairs, David E. Housman, PhD, Stephen J. Tapscott, MD PhD, and Silvere van der Maarel, PhD; organizer Daniel Paul Perez; all the speakers; and the meeting sponsors: Association Française contre les Myopathies,

*Where is all that DUX4 protein coming from? In 2012, Alexandra Belayew and her colleagues at the University of Mons-Hainaut in Belgium proposed that DUX4 protein could diffuse from one nucleus to others.*



or tissue samples are vitally important. Jeffrey Statland, MD, and his colleagues at the University of Rochester analyzed blood serum from 22 FSHD patients and 22 matched controls for the presence of 243 proteins. They found that 21 of the proteins were significantly increased or decreased in FSHD. "Markers for inflammation and

Cytokines, the FSH Society, the FSHD Global Research Foundation, Genzyme, the Muscular Dystrophy Association, the NIH Eunice Kennedy Shriver NICHD Senator Paul D. Wellstone MDCRC for FSHD at the University of Massachusetts Medical School, Quest Diagnostics/Athena Diagnostics, and Regeneron. [FSH Watch](#)



## Raising Awareness Through Football

English team proudly wears FSH Society logo

by **CRAIG MONKS**  
Worcester, England

**M**any people would recognize Worcester, in the United Kingdom, as the birthplace of Lea & Perrins® Worcestershire sauce. Others marvel at the beauty of the magnificent Worcester Cathedral, whilst some consider Worcester porcelain as some of the finest available. Few know that it is also home to one of the oldest football grounds in the U.K.!

For over 108 years, Worcester City Football Club has been sited at St. Georges Lane in the center of Worcester. Unfortunately, this year will be the last before it moves to a new home in 2015. However, ensuring the club has a solid foundation has been an absolute priority in the twilight years of the ground. To that end, Worcester City can boast a superb youth team group with ages ranging from under 12 to under 18. All of the age groups have been very successful in the current season and have fared very well in the leagues in which they play. Many of the young players will eventually play for the first team.

The under-12 age group is managed by local businessman Andy Twigg, whose business, Nutz4Sportz, supplies all of the kits used by the club. Andy has brought together a very fine group of young men whose talent on the pitch is matched by their gentlemanly conduct off the pitch and their desire to look after each other and form a footballing family. The 15 boys who form the squad have only lost two matches in the whole season, and their hard work on and off the pitch inspires pride from their coaches and families.

Recently, the age group was entered into a prestigious tournament in the southwest of England. As is normal for sports tours, it was decided that all of the team should be given a useful piece of clothing to wear and to create identity beyond the playing kits. To this end, each boy was given a high-quality black hoodie embellished with the club badge, details of the tour, and his initials. This, then, left a space on the hoodie that we felt should be taken up by something meaningful and selected by the boys and their parents.

One of our footballing family is Jacob Saul. Jacob's mother Trisha has FSHD. She is an inspiration to us all with her constant enthusiasm for the boys' football. With a wish to support Trisha in her ongoing battle with FSHD, the team unanimously chose to support the FSH Society, and with the kind permission of June Kinoshita, the tour clothing was emblazoned with the Society's logo.

The team hopes to adopt the FSH Society as our ongoing charity. Next season we hope that we will not only carry the FSH logo on our



**The Worcester City under-12 football team. Trish is seated in the middle. Her son Jacob kneels to her left.**

clothing, but also be able to undertake some fundraising. Knowing how much this would be appreciated by Trisha, Jacob, and their family, let alone others with FSHD, will be an inspiration to us all as our footballing family grows.

To give us all a greater insight and understanding, Trisha has very kindly shared her personal story with us.

### Trisha's Story

I was about seven years old when I remember being told to smile properly for the photo. I thought I was! Apparently, my smile was always crooked, and I think people thought I was pulling a face on purpose, but this was me at the very start of my journey living with FSHD. From then on I would do everything possible to avoid family photographs. People would often say to me, Why do you look so sad?, or Smile, it may never happen. This I know now was because of the lack of my facial expression, but as a little girl it made me become very shy.

I was 19 years old when I suddenly noticed I could no longer raise my left arm above shoulder height. I thought I had been a little too keen with my aerobics class, or that having started my general nurse training, I had pulled it whilst lifting a patient.

Following a visit to my doctor, I was given an urgent referral to a neurologist, Dr. Williams at The Queen Elizabeth Hospital, Birmingham. Thinking nothing serious was wrong, I went to the appointment on my own. After a simple examination, I was told I had muscular dystrophy.

I don't remember asking any questions, and I don't remember the journey back home. I was in complete shock. The only muscular dystrophy I was familiar with was Duchenne, and I knew this was life limiting. I thought I was going to die! On the way home I met my mum. She instantly knew something was wrong; I could hardly speak to her. Soon after, I had a series of blood tests and an EMG, and my initial diagnosis was confirmed.

I researched FSHD and found that in those early days, information often was quite conflicting, but I knew this was one of the mildest of the muscular dystrophy groups, and so I got on and lived life to the full—well, as much as this disease would allow me to.

I spoke to my nurse tutor who was extremely supportive, and I completed my nurse training in 1987—a very proud moment of my life.

No further involvement occurred until I was 28 years old, when my right shoulder became affected. Also at this time, I lost the use of my right bicep muscle. It now became increasingly difficult to lift items down from a height or to reach up for things. However, I have always been very independent and so would do things in my own way and only ask for help if I was struggling. This was my way of adapting to new situations.

This was also the year I got married to John, my boyfriend of seven years. John knew about my FSHD from very early on in our relationship and has always been my rock. He is there to comfort me when I have my bad days, yet treats me normally, and this is what I love about him so much. I think this is what has kept me so positive and active for all these years.

Both John and I loved and wanted children, and so had genetic counseling. My parents, two brothers, and sister were physically examined, and none were found to have FSHD, and so it was said that I was a mutation. Initially, we were told that this would not be passed on to any offspring and that I would only have wasting of my upper limbs. We were both elated, but this was short lived as it was then confirmed that I had a one-in-two chance of passing it on to our children and that there was the risk of my legs becoming involved.

We explored the option of adoption but were met by the stigma that we would not be eligible to adopt a healthy child. We decided we would take our chances, and we became the proud parents of Joshua in April 1996. (Prenatal testing was in its infancy at this time.) FSHD did not affect me during my pregnancy. In fact, I felt well throughout it and only had the normal pregnancy-related symptoms.

Shortly after having Joshua, my left leg began to feel different. This may sound strange, but this is how it felt. I now was beginning to have to put some effort into standing up from a crouched position. We decided to try for

another baby, but this time decided on prenatal testing so that we would know what to expect. I had chorionic villus sampling at 10 weeks of gestation. I was nearly 17 weeks pregnant when we were told that our second son (Jacob) was not affected.

By now I could no longer go up or down the stairs without holding a rail, and my legs were starting to become more unsteady. Therefore, I never carried Jacob in my arms when out of the house for fear of dropping him. As a mum, I often struggled with this. I had two beautiful young boys, and I couldn't pick them up if they fell over. I couldn't chase after them in a park or go for lovely strolls in the countryside.

For me it has always been the little things that have hurt so much, but I knew I had to stop this and turn the negatives into positives and concentrate on being a mum who could still do things like read stories, play games, and give lots of cuddles.

My legs continued to deteriorate. I began to wear a splint on my left foot for foot drop about seven years ago. I am now 48 years old, and I can only lift both of my arms approximately 30 to 40 degrees from my side. I walk with a splint and a walking stick for small distances, and I am now accepting that if I don't want to shut the many doors open to me, I will have to start using a wheelchair to remain active.

I use my wheelchair now for shopping and watching my son Jacob, who plays both football and rugby, both of which I love. I have come to understand that children want you there to support them and to encourage them, and that it doesn't matter if you are not like the other mums and dads.

I have continued to work. I fulfilled my dream of becoming a pediatric nurse. I have worked in a high-dependency/intensive care unit for neurosurgery and neurology. I have worked on a pediatric hematology and oncology ward at Birmingham Children's Hospital where I have specialized and now job share the role as the senior nurse

specialist for pediatric stem cell transplant services, of which Birmingham is the third largest center in the U.K. I plan and coordinate all West Midland transplants, provide transplant discussions as part of the consent process, and operate an apheresis machine, which is used for the collection of stem cells and other apheresis procedures. All this, however, is only possible because I have a very supportive team and manager who believe that you can still be a valuable member of the team despite your disability.

My message is simple: You can have a very rich and fulfilled life with FSHD, but you have to try to remain positive and adapt to the changes you have no control over. It is okay to feel sad sometimes and to long for what you can't have, but then you have to get up the next day and make a difference. [FSH Watch](#)



The Worcester City football hoodie



## My Story of Hope

*On never giving up*

by **PHIL BENNETT**

Nashville, Tennessee

I first started noticing muscle weakness in my shoulders when I was 16 or 17 years old. Until then, I thought I was just a normal teen. However, I never understood why I couldn't run as fast as the other guys or why I couldn't do sit-ups or pull-ups. Kids made fun of me because I ran so slowly. I just assumed I was a weakling and needed to exercise more.

For most, FSHD progresses so gradually that you don't realize you have lost another muscle until you go to do something and realize you can't do it anymore. When I was in my early twenties, I loved to hunt, especially bow hunt. I loved archery and was actually a pretty good shot. One early morning I was out in the woods and finally called up a couple of turkeys.

Now, wild turkeys are the most challenging to hunt, and I always wanted to get one with my bow and arrow. Here was my chance. When I thought the birds were close enough, I raised up my compound bow, but I couldn't pull it back. I did not realize until that moment that the shoulder muscles required to pull back the bowstring were now gone. That ended my archery career.

The same thing happened when I joined a work softball team in my mid-twenties. I loved to play softball and was pretty good at it when I was younger. Well, I went to our first practice, got up to bat, and hit the ball to right field. I was excited because I wanted to do well, but when I took off to first I realized that something was wrong. I was really having a hard time running.

Well, I got thrown out at first base by the right fielder. Now, guys on a softball team can be brutal, and I found that out immediately. I pretended to have hurt myself because at that point in my life, I was embarrassed and had no clue what was going on with my leg muscles. That ended my softball career.

So with two things I loved to do out of the picture, I decided it was time to see a doctor. I started with my family doctor. He referred me to an orthopedic specialist, who referred me to a neuromuscular clinic. I finally got an appointment, only to be poked, prodded, stabbed, twisted, and turned inside out without a diagnosis. They needed



**Phil Bennett**

to run more tests, but they thought I had some form of muscular dystrophy that they had not seen before. I was not in the mood for more tests!

I'll back up a bit and give you some family history. My Grandpa on my mother's side had similar issues, but never went to the doctor. My uncle on my mother's side has similar muscle weakness as well.

"You must have what Grandpa had" is all I ever heard from my family when I started having problems. Well, I did not accept that as a diagnosis and was bound and determined to find out what was wrong and hopefully get it fixed.

It wasn't until I was in my late twenties that I finally found a doctor in St. Louis who told me what I had. He said most likely I had facioscapulohumeral muscular dystrophy. I learned that they did not know what caused it, and there was no treatment at all.

So I just learned to live with it like Grandpa did and my Uncle Richard does. I was fine! Who needs to run anyway? Who needs to shoot a bow and arrow anyway? At least I was healthy and could still play golf, right? At least I can still play guitar and play the drums ... right?

In my thirties, I started noticing weakness in my abdomen, forearms, legs, and feet. I could not raise my arms over my head anymore. Climbing stairs became harder and harder. I started tripping and falling a lot. The problem was, once I tripped and

fell down, I could not get up off the floor without the help of a chair or solid surface. Also, the muscles in my abdomen weakened to the point where it looked like I was pregnant. Meanwhile, I stayed positive and hoped that someday researchers could find out what the hell caused this disease and give me some sort of hope for a treatment.

In my forties, I noticed the progression had slowed considerably. Even though I can barely climb stairs, cannot play my beloved game of golf, run or play ball, I am very blessed. I still play guitar even though muscles in my hands and forearms cause problems. I pretty much gave up playing the drums, mainly because it's too hard to carry all the equipment.

I am blessed!! Even though my disease has brought me to the point where I cannot continue working as a medical equipment technician in a hospital, I never give up.

A few months ago, I made the difficult decision to apply for disability and was approved. Luckily, my company offered short-term disability. After using up all my personal days and having almost run out of company-sponsored short-term disability benefits, I got a phone call from Human Resources. They asked me if I would like to interview for a job as a diagnostic cardiology technical support engineer.

Now this is the job I had been trying to get for three years! Four interviews and two months later, I landed the job. My start date was the exact date that my short-term disability benefits were set to run out. I work from a home office, no commute, nice raise, no tripping and falling in the halls of the hospital, no people staring at me wondering what is wrong. My dream job. I am so glad I never lost hope and for the good support I received from others.

You see, I have fallen down so many times in my life, but I always got back up. If you give up mentally, your body gives up as well.

I will never let my muscles tell me I can't play the guitar. That is one love I will never release. These days I find myself in awe of what can happen if you keep the faith and never lose hope.



## Celebrity Walk 'n' Roll 2013

If you have read this far, I'm impressed! I would like to tell you about a great organization, the FSH Society. Researchers funded by the FSH Society, as well as by other organizations like the National Institutes of Health and the Muscular Dystrophy Association, have made great strides in the last 10 years.

They now know what gene or set of genes is causing the problem. They have developed mouse models for treatment research. Things are looking good for a treatment or a cure in the next 10 years.

The FSH Society is a wonderful charity. They deal directly with the teams of researchers and fund their research directly; no middle people with their hands out. With very low administrative costs, virtually every cent goes to research, education, and patient services. This is impressive.

My current goal is to start raising funds for research. I am organizing a fundraiser here near Nashville, a night of music on February 26 (see below). Drop me a line at [songmaker11@gmail.com](mailto:songmaker11@gmail.com) if you would like to help.

Thank you for reading my story. Like Dr. Wayne Dyer says, "Don't let your music die with you." Never lose hope. [FSHWatch](#)

### Mark your calendars!

**Phil's Jam for FSH**  
February 26, 2014  
6:00 p.m.-11:00 p.m.

Enjoy an evening of music with some of the finest singer-songwriters in the Nashville area to benefit the FSH Society. The first annual Phil's Jam for FSH will be held at the Wild Wing Café in historic Franklin, Tennessee, located 14 miles south of Nashville. Several local songwriters will perform their hits and future hits. The Anna Johnson Band will perform, followed by Maureen and the Machine. There will also be a silent auction with all proceeds benefiting the FSH Society. Wild Wing Café offers a full menu with a portion of their sales during the event going to the FSH Society. \$5 donation at the door. Check the FSH Society website for further event details.



Walkers set out in Heritage Park, led by (from left) Obba Babatunde, Fabie Kay Combs, Millena Gay, Lydia Cornell, Melissa Biggs, and Roger Clark. Following behind (second row, left) in the Panama hat is Max Adler, who hosted the event.



Love birds! Cyndi Segroves flew in from Arizona to meet up with her fiancé Ray Jordan, who arrived from Australia.



Music legend Florence LaRue, of the multi-Grammy-winning group The 5th Dimension, treated walkers to a beautiful song.



# An FSHD Family Chronicle: Daniel's Story

Part 1 of the FSHD Guzik family chronicle

by **TIM GUZIK**  
Crescent City, California



**Daniel Guzik**

My father Samuel Guzik served in the U.S. Army 82nd Airborne Division during World War II, met our mother Frances, a Navy Wave, fell in love and married. They had 13 children together, eight brothers and five beautiful sisters.

Our parents taught each of us the importance of always doing our best, perseverance, resiliency, obedience, and most importantly, tolerance. They had no way of knowing that these simple values would be put to the ultimate test when four of their

children would be diagnosed with FSHD. Our brothers Dan, Tom, and Greg, and our sister Clarice were stricken with this insidious disease. Our parents never gave up hope that a cure would come.

A shooting star named Daniel streaked into our lives on August 17, 1950. His light and energy would fill the hearts of everyone that his life touched. Dan was my older brother, mentor, life guide, and hero.

From the beginning of his life, my parents knew there was something special about Daniel. He progressed normally until the age of six, when he began to exhibit the signs of FSHD: facial weakness, rolled shoulders, arched back, and feet that turned inward. A muscle biopsy was taken when Dan was about nine, and we finally had a diagnosis. It was FSHD. There were frantic attempts at therapy, but the relentless progression of the disease continued. By the age of 12, Dan was in a wheelchair.

Dan was raised with 12 brothers and sisters and was expected to pull his weight whether it was performing household chores, playing sports, doing well in school, or taking pride in his appearance. Dan was just one member of a large family, and he had to struggle for recognition and success just like all of the other siblings.

Dan developed the ability to crawl on his hands and knees and was rarely in his wheelchair. He had a very competitive spirit, especially when it came to sports. There were the grueling Friday night tackle football games in the living room while our unsuspecting parents were out grocery shopping. During basketball season, we played spirited games on the floor using a trashcan and a plastic basketball.

We played baseball on the driveway. Dan

While our parents waited and prayed for a cure, they were determined to treat each of their children equally and maintain the same level of expectation regardless of the special challenges that FSHD creates.

Our siblings with FSHD were given household chores that were suitable to their specific challenges. For example, Dan was required to dust furniture, clean baseboards, and vacuum. Each child was given appropriate expectations at home and in school, and then held to a high standard of performance.

Our family's emphasis on ability instead of disability created an atmosphere of mutual respect, achievement, and strength that endured throughout our lives. Education was of particular importance to our parents and resulted in seven bachelor's degrees, three master's degrees, and five associate of arts degrees. All of the kids with FSHD earned college degrees and held jobs within their disciplines.



**Daniel Guzik (right) as a child.**

pitched using a plastic ball while sitting on the ground and batted with a plastic bat. I was the designated runner, and if I didn't beat out the hit to first base there was hell to pay.

On Sundays, all of the brothers would go to the local park to play football. We played against anyone who came to the park to challenge us. Daniel was our quarterback. He would crawl up and down the field and throw from a sitting position. Woe to the receiver who dropped one of his passes; he'd have to face Dan's anger in the huddle. Dan was treated like any other player and got hit hard on many occasions. None of the brothers saw him as disabled. He was our quarterback, and he had better lead us to victory.

As we grew into our teens, Dan developed a strong personality and a fiery spirit. I was his younger brother, and I was supposed to channel Dan's love of sports as an athlete, but I fell far short of his expectations.

Dan was determined to work in the sports field as an adult to satisfy his competitive nature. After high school, Daniel enrolled in college and majored in sports information at California State University, Fullerton. He became a pioneer in overcoming the hurdles of attending college. This was before laws were passed requiring adaptations to buildings.

Dan was one of the founding members of the Handicapped Student Center on campus and threw parties at our apartment that are legendary. He arranged several fun trips to Hawaii and Florida before it was routine for people with disabilities to travel. As part of his studies, he became involved with the Los Angeles Rams and conducted interviews with the players and coaches.

Dan's interest in sports led him to contact the broadcasters of the Los Angeles Angels. One of the broadcasters, Don Wells, came to our house so that Dan could talk to him about the possibility of becoming a broadcaster. While at the house, Wells found out that our family were rabid Detroit Tigers fans, so he gave Dan box seats at the next Tigers game. Before the game, Wells met us, took us to meet the owner Gene Autry, and then to the dugout to meet our hero, now Hall of Famer Al Kaline.

Upon graduating from college, Dan was hired by the university as the Assistant Sports Information Director. He loved his job, his friends, and, most importantly, his family. Nothing seemed impossible for Daniel until a tragic car accident while traveling in Washington killed one of his friends and nearly



## Portraits of FSHD

A photo shoot in Atlanta

by BRENT ROWLETT  
Atlanta, Georgia

killed him. When my family received the call, my mother, father, and I flew to the little town of Port Townsend, Washington. We didn't expect Dan to still be alive when we arrived at the hospital. He was in extremely critical condition with multiple injuries including brain trauma that was causing his brain to swell. I took his hand in mine and whispered into his ear, and a tear fell from his eye.

The doctors didn't expect Dan to live, but they didn't know his spirit and determination. A few days later Dan began making strange motions with his fingers on his chest. We were puzzled but kept trying to understand what he was trying to do. Finally, an epiphany. Dan was drawing letters on his chest to spell words of love and comfort that he was going to be all right.

Dan did survive. In fact, he came back in ways that no one could have ever anticipated. There were many setbacks and two years of therapy, but slowly the Dan we knew re-emerged. The biggest loss was to his voice. Weeks of being on a ventilator reduced his voice to a whisper and with that, his chances of becoming a broadcaster.

After the accident, Dan was determined to regain his independence and live his life to its fullest. He placed an ad in the local paper for an aide and interviewed a young woman whose name was Jean. Jean said that she fell in love with Daniel during the interview, and a year later they were married. She later told me that she had never met someone with Daniel's charisma, strength, and wit, and couldn't help but fall in love with him.

Daniel and Jean were blessed with several years of marriage. Even though my brother's disease continued its relentless progression, their love only grew stronger. And then, just after attending my parents' 65th birthday party, my dear brother went to sleep at age 39, and his star returned to its resting place in the night sky. Daniel's life was cut short by the weakness in breathing caused by FSHD. His life was brief like the star that brought him to us, but his energy and light still burn brightly in our hearts. [FSHWatch](#)

**Editor's note:** You may have seen the photographs and stories of "Lives Affected by FSHD" in our year-end letter. This is the backstory by the photographer who created those images.

**M**y wife, Mary, and I were first made aware of FSHD through Christiane Wyckoff and her family. When high school senior year pictures were coming due for their daughter Carden, Christiane asked if I would be willing to take Carden's portrait in the comfort of our home.

Mary and I worked with Christiane and Carden to produce a stunning artistic portrait, a keepsake of which the family could be proud. The portrait was truly beautiful. Mary and I were thrilled that both Carden and Christiane were happy with the results.

When the FSH Society's Executive Director June Kinoshita asked Christiane to recommend a photographer to shoot portraits of FSHD patients at the Fulmer Family Dinner in September, I was honored that Christiane recommended me.

I was briefed by June that the purpose of the photography was to honestly portray the impact of FSHD on people's lives, but I had not anticipated the variety of portrayals that would be requested, which required constant movement of all the lights, but everything turned out well. My personal goal was to produce a warm keepsake portrait for each participant.

All of us are self-conscious about some part of our bodies when sitting for a photo. I was inspired by the courage of each participant to put those feelings aside and to display difficulties with movement and condition for others to view and be made aware of. We quickly bonded with all the participants, and I hope we gave them images with which they could be happy.

The image of Lester Fulmer comes to mind. He was so proud of his colorful Hawaiian shirt and lei. He deftly maneuvered his wheelchair right into position for his pictures. This was his fun night out, and we were happy to be a part. The way we posed Lester with the lighting makes a stately image worthy of display above a fireplace mantle.

We love every one of the people we met. Mary and I delayed celebrating our 34th wedding anniversary to do this special photo shoot for the FSH Society and Christiane. The warm memories of all these people are something we will not forget, a wonderful gift to us. Thank you. [FSHWatch](#)



Portrait of Lester Fulmer by Brent Rowlett

### PHOTOGRAPHER BRENT ROWLETT

Photographer Brent Rowlett is retired from a 30-year career in flexible packaging sales, marketing, product photography, and design with Fortune 500 companies Reynolds Metals Company and Crown Zellerbach Corporation. Operating his photography and design business, Eye Photo Design, Brent continues to design packaging, including food photography, and enjoys doing all types of event and portrait photography in the Atlanta area.

With extensive graphic skills utilizing the latest software, Eye Photo Design can produce not only the photography organizations require, but also create the packaging materials and printed publications to market their products and activities. Originally from San Diego, Brent and his wife, Mary, and daughters, Carrie and Cassi, have made Atlanta their home since 1983.

## Putting FSHD on the Run

At the NYC Marathon and Nike Half Marathon

by JUNE KINOSHITA  
FSH Society

Jeff Johnston's niece Becky Bridges and good friend Geoff Bello ran in the 2013 New York City Marathon to raise money and awareness for FSH muscular dystrophy and the FSH Society. This was Geoff's fifth year running the NYC Marathon for the FSH Society and Becky's second attempt. Last year, Geoff and Becky had to postpone their marathon effort because of Hurricane Sandy. This year's NYC Marathon runs raised \$13,600, for a tally of more than \$53,000 over the years.

In San Francisco, Paula Birnbaum ran the Nike Half Marathon to raise funds for the FSH Society in honor of her mother Barbara Birnbaum, who passed away in 2011 after a 17-year battle with FSHD.

"My family has chosen to channel our fundraising efforts through the FSH Society," said Birnbaum, "because it harnesses the power and insight of a patient-driven model to not only support those afflicted with the disease, as well as their families, but also to educate, advocate, and fund the necessary research to ultimately find a cure for the disease." Paula's run raised nearly \$4,000.

The FSH Society is truly grateful to Becky, Geoff, and Paula for committing their time and energy to train and run for these demanding races.

You can launch your own "run against FSHD" by contacting the FSH Society and setting up a fundraising page on Razoo. Just go to [razoo.com](http://razoo.com), search for "FSH Society," click on the FSH Society logo, and find, on the right side of that page, "Fundraise for this cause." Click the "Get Started" button just below that. Then start sharing your campaign with friends, and email [june.kinoshita@fshsociety.org](mailto:june.kinoshita@fshsociety.org) to let us know about your campaign! [FSHWatch](#)

# razoo



### RAZOO ONLINE FUNDRAISING

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

<http://www.razoo.com/story/Facioscapulohumeral-Society>

### 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!

### DOES THE SOCIETY HAVE YOUR CURRENT ADDRESS?

Be sure to receive breaking news and other up-to-the-minute information from the Society. Please send us your address to [info@fshsociety.org](mailto:info@fshsociety.org) or drop us a line at:

FSH Society  
450 Bedford Street  
Lexington, MA 02420



Don't forget to include your email address and phone number. Thank you!



Geoff Bello's marathon jersey honors his friend Jeff Johnston.



Paula Birnbaum, center

## The Fourth Annual Fulmer Family Dinner

*Bringing together FSHD folks from all over*

by **ROD FULMER**  
McDonough, Georgia

**F**our years ago when I started trying to come up with an idea of how to raise money for FSHD and a group of people whom we have all come to know as the FSH Society, I thought it would be kind of neat to have an evening where people could come together to enjoy some great food, live entertainment, and maybe win a door prize.

We were not sure that people would even attend. That first year we had a little more than 50 people and raised about \$1,800. This year, we had more than 150 people and raised about \$10,500!

Looking back at this year's dinner, I am pleased at the amount raised. Even more than that, the thing that comforts me the most is that we had so many people with FSHD attend.

One of my desires when we started these dinners was to pull people with FSHD together and have an evening where we could talk and visit. Thanks to the FSH Society and one of our Atlanta doctors, people have found out about the dinner and now know that there are others in the Southeast who share the same concerns, desires, and hopes in life.

I know there are other fundraisers for the Society that raise far more money, and I can't dwell on that. The thing that I care about is bringing people together, and who knows which dollar and where it was raised that will put us over the top and allow a cure or treatment to be found?

I have so many people to thank who have worked so hard along the way, including my wife and daughter, my mom, our extended family, and some of our dearest friends. The greatest compliments were from those whom we have known for years who attended for the first time this year. They came up to us with tears in their eyes and said they wished they had been coming all along. [FSH Watch](#)



Gracing the Fulmer Dinner this year were the award-winning Peach State Quartet and Virginia Wyckoff, 2013 Miss Georgia Teen.



## Waiting for Manolo

*Adapting to my AFOs*

by **ELLEN HANNAN**  
Old Greenwich, Connecticut



**A**t first, I thought it was my shoes. Late on a Friday, after a long day in a long week of meeting clients in several cities, I found myself trying to run for a plane in the Dallas airport. Despite having swapped my stylish heels for sensible flats, I ended up taking off my right shoe and racing, half-barefooted, determined to arrive at the gate before the plane's doors were closed. I made the flight, but a pattern was forming. New shoes, with ever lower heels seemed to be required with increased frequency.

Several years later, I exited a cab late one rainy night in New York City. A city bus loomed in the distance a block or two to my south, safe enough to make a dash to the other side of the street. While I made it across, my right foot was now of no help at all. Although I had long since segued to flats, my Manolos and Jimmy Choos continued to gather dust in my closet waiting for me to find a way to make them work for me again.

Fast-forward several more years to when I received my diagnosis of FSHD. The Muscular Dystrophy Center at White Plains Hospital in New York recommended a physical therapist well versed in FSHD. He finally convinced me that an ankle-foot orthosis (AFO) was the way to go, as my gait was beginning to compromise my hip and knee joints, and it was no longer just about the stylish footwear.

I'll admit to being quite unprepared for the alternatives available—particularly the shoes that came along with the leg braces. However, I was fortunate to find an orthotist (someone trained in the assessment and fitting of AFOs) who was sympathetic to my needs and recommended the brand that I settled on, the Otto Bock Walk On Trimable.

A cautionary note is warranted. My particular weakness is in the lower leg muscles that control the foot flexors. My hamstrings and quads are still relatively strong. Thus, while this particular design works well in offsetting a foot drop weakness, it may not be as helpful in compensating for upper leg weakness.

My suggestion for those who may be considering AFOs: Don't settle for the first model you see. Ask questions, try different models, and watch for new research using ever lighter materials. Surely, something newer and better is on the horizon!

I still hope that someday Manolo Blahnik will bring his not inconsiderable talents to bear on designing a beautiful shoe that will fit over my AFOs. In the meantime, while we search for a cure for FSHD, I can still manage a set of stairs. I'm not as graceful as I'd like, but I'm grateful for the additional mobility my AFOs provide. [FSH Watch](#)

# An Enchanted Evening

*Benefit concert smashes record*

by **BETH JOHNSTON**  
Denver, Colorado

The 2013 Festive Evening of Song was held on September 30 at the New York Botanical Garden. This wonderful event to benefit the FSH Society and FSH muscular dystrophy research was attended by nearly 300 guests. The evening was magical, complete with an elegant dinner buffet, a surprise visit and greeting from actor Max Adler, and a remarkable concert performance by pianist Steven Blier and multi-Tony Award-winning singer Judy Kaye.

Raising nearly \$320,000, the event accomplished its goals of creating awareness of the disease and of the FSH Society and raising critical dollars for FSHD research. Co-chairing the event were Judy Seslowe and Beth Johnston. A special thank you to Sheila Cohen and the entire Concert Committee, who worked tirelessly to ensure such success; to Bob and Abigail Kirsch for providing the gracious dinner and event venue; to Max Adler, who has given and continues to give his full support to the FSH Society; to Bill Milling and Susan Egert-Milling, who wrote and produced our latest public awareness video; and, of course, to Steven Blier and Judy Kaye, who shared their amazing musical talents with this lucky audience. [FSH Watch](#)



Broadway legend Judy Kaye entertained the audience with her soaring vocals and impeccable comic timing.



From left, Takako Jones, Peter Jones, Daniel Paul Perez, Susan Perez, and Jennifer Egert.



From left, Beth Johnston, June Kinoshita, Max Adler, and Jennifer Bronstein.



Concert co-chairs Judy Seslowe and Beth Johnston.



Concertgoers enjoyed an elegant dinner at the New York Botanical Garden.

## GET SOCIAL!



Join our online communities to get news, ask questions, and seek advice and support from fellow FSHD patients and family members. The FSH Society Yahoo! Groups forum has tens of thousands of searchable posts. Bookmark these pages and come back often. To find the FSH Society Facebook page and Yahoo! Groups, go to our homepage at <http://www.fshsociety.org>, click on the "Community & Reference" menu tab at the top of the page, and select "Online Community" in the left-side 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.



## MATCHING GIFTS AND OTHER WORKPLACE GIVING

You may have an opportunity to support the FSH Society this fall when you make a United Way pledge for 2014. Check with your Human Resources department at work for more information.

## CHARITY NAVIGATOR TOP PERFORMER

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



[www.charitynavigator.org](http://www.charitynavigator.org)