

# FSH Watch



CONNECTING THE COMMUNITY OF PATIENTS, FAMILIES, CLINICIANS AND INVESTIGATORS

 LIVING WITH FSHD

## FSHD and my journey of faith

by DEBORAH CALHOUN

I was diagnosed with FSHD Muscular Dystrophy at the age of 10. Receiving this news at such a young age means I have few strong memories of life “before” the diagnosis; it means I don’t have one significant moment of knowing my life had changed, the type of moment Eric Lowen so poignantly describes in his song “How Mighty is the Silence” of when he learned he had a neurodegenerative disease. In fact, I can’t imagine who I might be today if FSHD hadn’t been a companion nearly all my life.

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Deb and her father, Ed Sullivan

## A Festive Evening of Music & Song raises over \$180,000 for FSHD research



Good friends make a great committee! Judy Seslowe, concert co-chair, and member of the FSH Society Board of Directors, in front. More photos on pages 8 and 9.

 LIVING WITH FSHD

## The decision to stop working

As their FSHD progresses, some people find that reduced stamina, continually increasing fatigue and intractable physical problems make it increasingly difficult to continue working. They want to continue working but “hit a wall” in their ability to do the job, and also don’t want to risk accelerating the progression of their FSHD by continuing to work past the point of prudence. The decision to stop working and “go on disability” is one of the most agonizingly difficult decisions one ever has to make—emotionally, practically and financially.

Making the right decision requires advice and support from one’s spouse, other family members, doctor, and other professionals who might include occupational therapists, social workers, psychologists or psychiatrists, and lawyers. Taking the first step can be especially difficult—talking candidly to your spouse, family and doctor about your difficulties at work.

Well before deciding to stop working, one must ask one’s employer for a “reasonable accommodation.” The Americans with Disabilities Act requires employers to provide a reasonable accommodation; depending on the circumstances, this could include an evaluation by an occupational therapist or workplace ergonomic expert, assistive technology, a change in job duties, and reduced hours. But as a person’s FSHD

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**LETTER FROM THE EXECUTIVE DIRECTOR**

Dear Friends,

FSH Society members, family and friends have contributed talent, energy and funds to events in the first months of 2010. Some of these events are reported within—they have themes of music and dancing, walking and rolling, as you will see, or as perhaps you have experienced when you attended one of these benefits. Other events are planned for later in the year.

When groups of volunteers form committees to help the Society, you often make the decision to designate the funds to FSHD research—funds that lead us closer to treatments and a cure. This issue of *Watch* also reports research results that have been made possible only through generous philanthropy, and investigators cite their appreciation for this financial assistance which you help to bring about.

But while we wait for therapies, life goes on, and many of you are living it well, for yourself and others. This issue also carries an example of an FSHD patient reaching out for others. And, there is also an article that discusses working and not working, to bring the full perspective.

Now, back to music and dance. Some would say, it is “perfect harmony,” a symphony of patients, friends, and our families working together to bring funds to support research and other work of the Society. At the same time, people affected with FSHD are doing their very best work for themselves and others.

All good wishes to you. We hope to see as many of you as possible at the FSH Society 2010 International Patient and Researcher Network Meeting, Las Vegas, Nevada, July 30 through August 1, 2010.

Sincerely,

Nancy Van Zant  
 Executive Director



**FSH Watch**

SPRING 2010

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It is the editorial policy to report on developments regarding FacioScapuloHumeral Muscular Dystrophy (FSHD), but not to endorse any of the drugs or treatments discussed. We urge you to consult with your own physician about the procedures mentioned.

The FSH Watch is published by the FSH Society and distributed by mail and electronically to its members and supporters.

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To be placed on the mailing list or to submit an article, please write to:

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Articles may be edited for space and clarity. Every effort has been made to ensure accuracy in the newsletter.

If you wish to correct an error, please write to the above address.

Look for us on the internet at: [www.fshsociety.org](http://www.fshsociety.org)

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## THE DECISION TO STOP WORKING

... from page 1

progresses, accommodations that once enabled him or her to work may no longer do so. One should contemplate going on disability only after having determined, together with one's employer, that further accommodations would not be feasible.

There are two types of disability insurance: Social Security disability insurance (SSDI), a program of the Social Security Administration, and long-term disability insurance (LTD). Everyone who works and pays Social Security taxes is automatically enrolled in SSDI. LTD policies are issued by insurance companies. A group LTD policy, where individual underwriting is not required, is often one of the most valuable benefits provided by employers. It may also be possible to enroll in a group LTD policy through a professional organization, alumni association, religious organization or other group. Individual LTD policies are also available but require individual underwriting, making it difficult for most people with FSHD to qualify. Since a group LTD policy does not require underwriting, people with FSHD who have an opportunity to obtain coverage under a group LTD policy are well advised to seize that opportunity. If one has an opportunity to purchase an individual policy because his FSHD symptoms are mild, this is also an opportunity to be seized. Because SSDI will not provide nearly as much income as working, supplementing Social Security insurance with a private LTD policy can significantly increase one's disability income.

Central to disability insurance is the definition of disability. SSDI has a strict, narrow definition of disability, known as an "any occupation" definition. Someone is not considered disabled if they merely cannot do their current job; they must not be able to perform any other job for which their education, training and experience would otherwise qualify them. The Social Security website has a step-by-step explanation of this definition: <http://www.ssa.gov/dibplan/dqualify4.htm>. Private LTD policies vary in their definition of disability, ranging from "any occupation" to a more liberal "own occupation" definition, which means the person is considered disabled if he is no longer able to perform his current occupation. For example, a truck driver may no longer be able to drive a truck but may be qualified by training and experience for office work; under an "any occupation" definition, he isn't disabled; while under an "own occupation" definition, he is.

The amount of the payment is critical. SSDI is based on average earnings over one's career, provides a modest fraction of the income earned while working, and has annual cost-of-living increases. Private LTD policies typically provide 60% or two-thirds of the income earned immediately before going on disability and generally do not provide inflation protection. Some private policies offset (subtract) the amount of Social Security disability income from the amount they pay; however, cost-of-living increases in

It is one of the biggest losses caused by our disease. Mental health professionals and social workers can greatly help those who are faced with the decision to stop working.

Before deciding to apply for disability, it is also advisable to get advice from a lawyer who is expert in disability insurance law. A lawyer should review your LTD policy and advise you on how to fill out the claim forms. Disability insurance law is a specialized field of law that is separate from the field of disability civil

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**“The decision to stop working and ‘go on disability’ is one of the most agonizingly difficult decisions one ever has to make—emotionally, practically and financially.”**

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Social Security disability income typically are not subtracted.

Another essential element is the duration of disability payments. SSDI payments continue until normal retirement age (65 or older, depending on the year of birth); thereafter, they are converted to regular Social Security retirement benefits in the same amount. Private LTD policies vary, ranging from payments for a specified number of years to payments through normal retirement age (usually 65).

It is impossible to overstate the importance of getting professional advice and support. The Social Security Administration and insurance company determinations (both initial and ongoing) about whether or not you are unable to work because of your FSHD will be based on a certification from your doctor and a review of your medical records. Having a doctor who knows you well and has been familiar with your FSHD for a long time is extremely helpful, both in helping you consider such a difficult decision and because he or she will have much greater credibility with the Social Security Administration and the insurance company than a doctor who has known you only a short time or who is not very knowledgeable about muscular dystrophy.

Working gives us creative fulfillment, intellectual stimulation, social camaraderie and a sense of accomplishment. It fills our waking hours and is an important part of our identity. Being forced to stop working because of FSHD is difficult emotionally and socially.

rights law. (The latter is concerned with discrimination in employment, physical access and government programs.) In fact, many lawyers who practice Social Security disability law are not familiar with private LTD insurance law, and vice versa, so if you have an LTD policy it may be necessary to consult with two lawyers. Your local bar association probably has a referral service where you can find lawyers with this expertise. Nolo, a publisher of self-help legal guides, publishes a guide to Social Security disability: [www.nolo.com](http://www.nolo.com). This book is a good place to start, but the fees for a couple of hours of legal consultation before filing a claim will be money well spent.

Taxes and medical insurance must also be considered. Federal income tax treatment of Social Security benefits is complex and depends on the overall level of income. In many states, Social Security benefits, including disability payments, are exempt from state income taxes. Generally, federal and state taxability of private LTD payments depends on who paid the insurance premium; payments are taxable if the employer paid the premium and tax exempt if the employee paid it. The IRS website, [www.irs.gov](http://www.irs.gov), has comprehensive information. See Publication 907 (Tax Highlights for Persons with Disabilities) and Publication 575 (Pension and Annuity Income).

Everyone who receives SSDI payments is automatically eligible for Medicare beginning 24 months after the date of disability.

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# Report from Paris: FSH Society helps renowned research institution make

by JULIE DUMONCEAUX, PH.D., and GILLIAN BUTLER-BROWNE, PH.D.  
Institut de Myologie, Université Pierre et Marie Curie, Paris, France

Our laboratory, which focuses on therapies for disorders of striated muscles, is part of the Institute of Myology, established by leaders, patients and families from the French Muscular Dystrophy Association (Association Française contre les Myopathies or AFM) to create a center for international expertise on neuromuscular diseases. The institute includes specialized clinical consultations, basic and clinical research, and teaching on muscle and muscle diseases. The institute is attached to and supported by Université Pierre et Marie Curie, National Institute of Health and Medical Research (Institut National pour la Santé et la Recherche Médicale, INSERM) and CNRS, both government-funded research organizations (equivalent to the U.S. National Institutes of Health or NIH), the Assistance Publique-Hôpitaux de Paris, and the Nuclear Energy Commission (Commissariat à l'Énergie Atomique). The Institute of Myology is currently involved in four of the six multicenter clinical trials in Europe. Our research laboratory, with approximately 100 people, is directed by Professor Thomas Voit. Its primary objective is to develop cell, gene and pharmacological therapies based on understanding the molecular physiopathology of several neuromuscular diseases. Programs are under development for Duchenne Muscular Dystrophy, Emery Dreyfuss Muscular Dystrophy, Myotonic Dystrophy, and Oculopharyngeal Muscular Dystrophy, and now we are adding FSHD.

Gillian Butler-Browne, Ph.D., is a research director employed by INSERM and is in charge of one of the research groups within this institute; Julie Dumonceaux, Ph.D., is a group leader employed directly by the Institute of Myology who has recently joined Dr. Butler-Browne's research group. One of the specialties of this group is human muscle cell culture. Vincent Mouly, Ph.D., in collaboration with Woodring E. Wright, MD, Ph.D. (University of Texas Southwestern Medical Center and the U.S. NIH Boston Biomedical Research Institute Sen. Paul Wellstone Cooperative Research Center for FSH Muscular Dystro-

phy), has recently developed a technique which enables them to generate non-tumorigenic immortalized cell lines from human skeletal muscle. This is a significant contribution because cells isolated from human muscle biopsies can divide only a limited number of times

Medical Center), has given us the opportunity to generate a very important resource for the community of FSHD researchers. We know that a high genetic heterogeneity exists among individuals. Heterogeneity dramatically complicates our experiments because it is impossible to determine if

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*“...access to funding specifically directed towards FSHD enables us to dedicate our efforts towards ... identifying new biomarkers and studying therapeutic approaches for FSHD.”*

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before they stop dividing and reach cellular death. Making new muscle cultures requires a regular supply of muscle biopsies from well characterised patients; this muscle tissue is not always available. To address this problem, our laboratories have developed an approach to immortalize muscle cells by transduction of two genes with viral vectors. Once incorporated into the muscle cells, these genes enable the cells to grow forever without losing their differentiation capacity. Interestingly, one of the first cell lines developed by Dr. Mouly was from an FSHD patient. This stimulated our interest in this pathology, and we have recently initiated a new research program focusing on FSHD.

FSHD is the most common muscular dystrophy and is caused by partial deletion of a repeated region of like DNA segments collectively called D4Z4 situated in the subtelomere of chromosome 4q in the majority of patients: healthy patients carry 11 to 100 copies of the D4Z4 repeat whereas FSHD patients have between one and 10 D4Z4 repetitions. It is unclear whether or not the deregulation of these genes is the cause or the consequence of the disease. Our laboratory will study this disease from a new angle, attempting to identify the role of a new class of RNA called microRNAs (miRNA) in the disease progression.

An interesting and fruitful collaboration with a leading geneticist on FSHD, Silvère van der Maarel, Ph.D. (Leiden University

the variation observed between healthy and FSHD samples is a specific characteristic of the FSHD pathology or is due to the genetic heterogeneity among the patients. Mosaicism denotes the presence of two populations of cells with different genotypes (the genetic makeup of an organism) within one individual who has initially developed from a single fertilized egg. These patients are unique since they do have the same genome in all their cells, apart from the presence of the mutation, with some cells carrying the D4Z4 deletion whereas the others do not. The percentage of mosaicism varies from one patient to another, depending on when the mutation has occurred during the embryologic development. The mosaic patients represent only a small percentage of all FSHD patients and, clinically, all patients have the same symptoms even though not all of their cells are affected. It is only when the genetic diagnosis is performed that the mosaic patients can be identified. Dr. van der Maarel has provided our lab with muscle biopsies from both affected and non-affected muscles of mosaic FSHD patients. We have been able to isolate immortalized lines with and without the D4Z4 deletion from these biopsies. These mosaic cell lines will allow us to bypass the complications caused by heterogeneity since healthy and diseased FSHD cells come from the same biopsy, the same environment, the same person, and have the same genetic

# inroads into FSHD

history. These cells are a valuable resource which will help us not only to obtain a better understanding of the disease but can also be used to develop and test new therapies for FSHD.

We are interested in microRNA expression levels in the mosaic cells because:

**1.** RNAs constitute one of the most important discoveries made in recent years in the field of molecular biology. They can precisely regulate the expression of a specific protein by inhibiting its translation and/or promoting the degradation of its mRNA [messenger RNA]. miRNAs are key regulators of gene expression and their misregulation in different disease progression/onset is an important consideration.

**2.** Several misregulations of genes have been shown in FSHD, suggesting a common mechanism between these deregulations. miRNAs represent an obvious candidate to explain these deregulations.

Our first effort is to evaluate the expression level of miRNAs in disease and healthy clones established from mosaic FSHD patients. To investigate the role of miRNA in the FSHD pathology, we will compare the expression level of different miRNAs in healthy and diseased immortalized cells, comparing as a control the expression between healthy immortalized and non-immortalized cells. We will next determine the effect of this modified expression: the miRNAs which are found to be up-regulated in the diseased cells will be artificially down-regulated and the miRNAs that are found to be down-regulated will be up-regulated. We will then look at the effect on the phenotype (the observable physical or biochemical characteristics of an organism, as determined by both genetic makeup and environmental influences) of the cells such as, in vitro, the ability of the cells to proliferate, or, in vivo, the ability of the cells to correctly make muscle by injecting them into the muscle of immunodeficient mice. This system allows us to evaluate the

myogenic potential of human cells in vivo by counting the number of human nuclei or the number of fibres expressing human proteins within the mouse muscle.

Thanks to the funding we obtained from the FSH Society, our laboratory has been able to continue this formative FSHD project. This financial support is crucial since the choice of projects carried out in the laboratory is driven by the funding available. Most government funding targets diseases such as cancer, Alzheimer's disease, cardiovascular disease and diabetes; access to funding specifically directed towards FSHD enables us to dedicate our efforts towards providing new insights into the pathology, identifying new biomarkers and studying therapeutic approaches for FSHD. We are also establishing a network with other researchers and, most importantly, geneticists and clinicians to pave the way towards these future therapeutic options.

This research is currently supported by the FSH Society Delta Railroad Construction Company Fellowship Grant.



## NEWS AND EVENTS

### International Patient and Researcher Network Meeting, July 30—August 1, 2010, Las Vegas

**S**ponsored by the FSH Society, the International Patient and Researcher Network Meeting is a partnership among patients, families, clinicians and scientists. It will include lectures by and question and answer sessions with leaders in the field of FSHD, reports from major clinical centers and research centers, and educational sessions on the genetics of FSHD, scapular fixation surgery, breathing and respiration, and exercise and physical therapy. Popular breakout sessions include topics that participants have requested as well as the interests of teenagers and young adults.

Overnight accommodations are available at Paris and Bally's Las Vegas. The FSH Society has a special conference rate of at \$89.00 (Bally's) per night (single or double occupancy) and \$120.00 (Paris) per night (single or double occupancy), plus taxes. These facilities have many wheelchair accessible guestrooms, including a total of 100 rooms with roll-in showers. There is no charge for parking. The group code for our conference rate for Bally's is SBIPRO

(last digit zero) and for Paris, SPIPRO (last digit zero). For the best selection of accessible rooms and showers, please make your reservations early. The closing date for the Society's block of rooms is July 9th. For reservations, call 1-877-603-4389, or reserve online

at <http://www.harrahs.com/CheckGroupAvailability.do?propCode=BLV&groupCode=SBIPRO> for Bally's, and <http://www.harrahs.com/CheckGroupAvailability.do?propCode=PLV&groupCode=SPIPRO> for Paris.

Registration for the meeting, including lunches Friday-Sunday, is \$180 per adult (with current FSH Society membership), \$200 for non members, \$105 per young adult age 12 – 18, and no charge for children under 12.

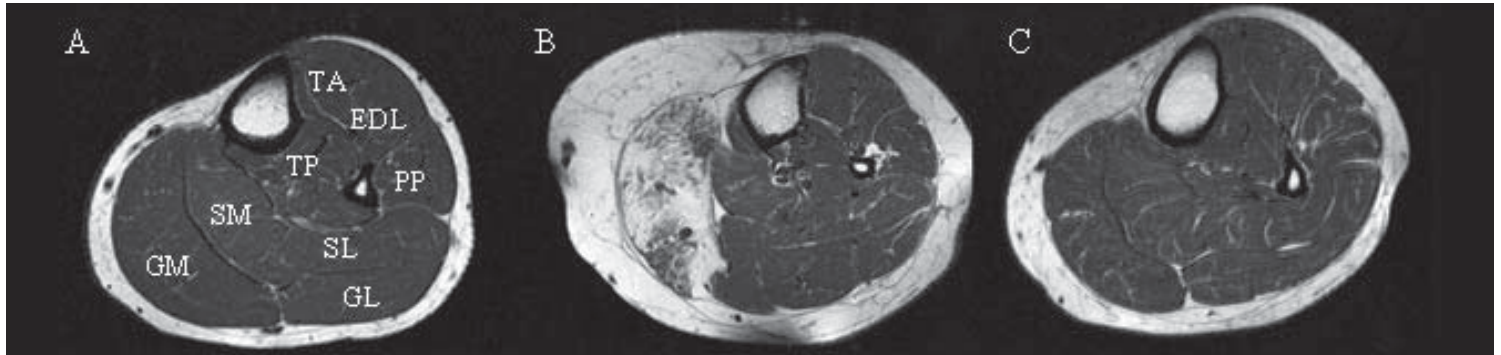
The full program and registration are available at [www.fshsociety.org](http://www.fshsociety.org), or e-mail [2010Meeting@fshsociety.org](mailto:2010Meeting@fshsociety.org). We hope to see you there!



# Sam E. and Mary F. Roberts Nutrition Research Fellowship Grant helps Dutch metabolic differences in FSHD muscle using MRI/MRS

Edited by **DANIEL PAUL PEREZ,**

President and CEO, FSH Society, from material received from Arend Heerschap, Ph.D., Radboud University Nijmegen Medical Center, Nijmegen, The Netherlands



MR (magnetic resonance) images of cross sections of the lower leg of a healthy volunteer (A) and two individuals with FSHD (B, C). In (B), the concentration of fat is visible in the gastrocnemius medialis muscle (GM), while other muscles appear to be normal. The second patient (C) shows no fat infiltration.

Important new research was published earlier this year showing that non-invasive imaging such as magnetic resonance imaging (MRI) and magnetic resonance spectroscopy (MRS) can be used to study the progression of FSHD. The work proposes several new potential non-invasive biomarkers such as metabolic differences between muscles with heavy fat content and muscles without heavy fat in FSHD patients, and levels of energy in muscle cell. The research was conducted at the Department of

Radiology and the Department of Neurology, Radboud University Nijmegen Medical Center, Nijmegen, The Netherlands. The research team was led by Hermien Kan, Ph.D., and Arend Heerschap, Ph.D.

This work was initiated and funded by the FSH Society Sam E. and Mary F. Roberts Nutrition Research Fellowship Grant. This pilot work subsequently picked up substantial funding from the Dutch government.

In carrying out this study, the Dutch

researchers have applied MRS to FSHD patients for the first time. The technique uses spectra, or radio waves, to measure the presence of various compounds in muscle. The spectra produce signals that record levels of energy in muscle cell—energy is the chemical capacity of a cell to function.

For the first time, researchers could measure spectra from several individual muscles separately. They observed abnormalities in FSHD muscles in the lower leg. For example, they found a decrease in what is called phosphocreatine, an important energy source in muscle, and a decrease in acidity. Furthermore, these changes were only observed in muscles that were fat infiltrated (see figure) as identified in the MR images in the FSHD patients, which suggest that in FSHD disease, progression is truly muscle specific. Non-fat infiltrated muscles in FSHD also had normal spectra and compounds.

The MRS observations point to an energy problem in the affected muscles. Similar abnormalities in phosphocreatine and pH have been observed for other muscular dystrophies and thus are not disease specific. In FSHD, the researchers did not find an increase in inorganic phosphate, which is commonly found in other dystrophies. This may possibly be considered as more disease specific. It also seems that the abnormalities correlate directly with the severity of affected muscles.

Dutch researchers will work with these findings and further investigate the cause

## FROM THE AUTHORS' ABSTRACT (see list of authors on page 7):

**"The aim of our study was to use a combination of <sup>31</sup>P magnetic resonance spectroscopic imaging (MRSI) and T1-weighted MRI to uncover metabolic differences in fat infiltrated and not fat infiltrated muscles in patients with FSHD. T1-weighted images and 3D <sup>31</sup>P MRSI were obtained from the calf muscles of nine patients with diagnosed FSHD and nine healthy age and sex matched volunteers. Muscles of patients were classified as fat infiltrated (PFM) and non fat-infiltrated (PNM) based on visual assessment of the MR images. Ratios of phosphocreatine (PCr), phosphodiester (PDE) and inorganic phosphate (Pi) over ATP and tissue pH were compared between PFM and PNM and the same muscles in healthy volunteers. Of all patients, seven showed moderate to severe fatty infiltration in one or more muscles. In these muscles, decreases in PCr/ATP and increases in tissue pH were observed compared to the same muscles in healthy volunteers. Interestingly, these differences were absent in the PNM group. Our data show that differences in metabolic ratios and tissue pH in skeletal muscle between healthy volunteers and patients with FSHD appear to be specific for fat infiltrated muscles. Normal appearing muscles on T1 weighted images of patients showed normal phosphoryl metabolism, which suggests that in FSHD disease progression is truly muscle specific."**

# investigators uncover

and origin of these abnormalities, e.g., by studying light exercise of the affected muscles using MRI. Further, they hope to use MRI to establish measurements and biomarkers to study how muscle goes from healthy to unhealthy. Such a scale of measures would enable the non-invasive assessment of the progression of disease using MRI. Finally, the technique of MRI might be used in the early stages of a long clinical trial to see early on whether a therapy is effective, rather than waiting months or years.

This is an interesting and unique paper that identifies several new non-invasive biomarkers. It also points to MRI/MRS as an important technique in the design of clinical trials, for documenting endpoints and measurements. The investigators, supported by the FSH Society and the Sam E. and Mary F. Roberts Fund, have made an important, innovative contribution to scientific understanding of FSHD.

To read the full scientific study, go to *"Only fat infiltrated muscles in resting lower leg of FSHD patients show disturbed energy metabolism,"* by Kan HE, Klomp DW, Wohlgemuth M, van Loosbroek-Wagemans I, van Engelen BG, Padberg GW, Heerschap A. Department of Radiology, Radboud University Nijmegen Medical Center, Nijmegen, The Netherlands. NMR Biomed. 2010 Feb 19.

The screenshot shows the FSH Society website. At the top, the logo for the FSH Society (Facioscapulohumeral Muscular Dystrophy) is visible. Below the logo is a navigation menu with links for Home, FSH Society, Contribute, About FSHD, For Patients, For Scientists, Research, Community & Reference, News & Events, and Contact. The main content area includes a large image of a shoulder, a 'Welcome to the FSH Society' message, and a 'Glad you found us! Come in.' section with a 'donate' button and the text 'Donate today!'. Below this is a 'Get Involved!' section with links for 'Have Questions? Call, E-mail, or Write Us!', 'Support Our Work, Make a Difference!', and 'Consider the gift of stock in 2010!'. A 'Newly Diagnosed?' section offers to talk to someone who understands. The right sidebar features a 'News & Events' section with a list of news items, each with a date and a brief description, such as 'The 2010 FSH Society International Research Consortium & Research Planning Meeting has been announced - Save the Date!' and 'Research pioneered by FSH Society leads to new discoveries using non-invasive MRI/MRS imaging of FSHD'.

**Did you know ... the Society recently updated the website. It is now easier to get the information you need on FSHD and keep up-to-date with the Society. Check it out!**

## THE DECISION TO STOP WORKING

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Medicare Part A (hospital insurance) is free for most people. Most people have to pay for Part B (medical insurance); premiums are deducted from monthly SSDI payments. Parts A and B do not cover everything, so most people also choose to get Medicare supplemental insurance (Medigap), which is provided by private insurance companies and paid for by the insured. Everyone on Medicare because he is 65 or older has an automatic right to purchase Medigap insurance from any company serving his geographic area. But the situation is different for those on Medicare because of disability. State law, not federal, governs whether or not private insurers are required to offer Medigap insurance for people who are on Medicare because of disability; some states require it and others do not.

The decision whether or not to stop working also involves complex considerations of medical insurance, especially from the last day of work until the beginning of eligibility for Medicare. Factors

include whether a continuation of your employer's insurance is available through COBRA, whether insurance through COBRA is affordable, and whether you can obtain medical insurance through your spouse's employer.

California Health Advocates, [www.cahealthadvocates.org](http://www.cahealthadvocates.org), and The Hawkins Center, [www.hawkinscenter.org](http://www.hawkinscenter.org), (510) 232-6611, provide comprehensive information about Medicare. Although these organizations are geared toward California consumers, much of the information applies to residents of all states.

*Editor's Note. This article is provided for general informational and educational purposes only. This article, and the FSH Society, do not provide medical, legal, financial or other advice or recommendations. Anyone considering filing a claim for disability should consult his or her professional advisors.*



## A Festive Evening of Music & Song raises over \$180,000 for FSHD research

Over 250 concert goers enjoyed a splendid evening to benefit the FSH Society on May 12, at the New York Botanical Garden, Bronx, New York. Steven Blier, co-founder and artistic director, New York Festival of Song, with John Brancy, baritone, played and sang a wonderful concert! Titled, "American Songbook," the concert featured the works of Cole Porter, Hoagy Carmichael, and Richard Rodgers. Steve delighted the concert goers with humor, good spirit and commentary in his introductions.

A silent auction and dinner preceded the concert and a dessert buffet followed it. Special thanks from all the FSH Society to these wonderful musicians, to Judy Seslowe and Beth Johnston, co-chairs of the concert; to Bob and Abigail Kirsch for enabling the use of the Garden Terrace Room at the Botanical Garden and to their fine staff for wonderful food and service, and to the concert committee for making this the most successful evening to date for the Society in New York.



From Steven Blier "I play many concerts all year, and each one assumes a tremendous amount of intensity for me. But these benefit performances that I have done to raise money for research on FSH muscular dystrophy have a special place in my heart. I cope with my disability on an hourly basis, but I rarely get a chance to do anything about it. What a joy to take up my own cause by playing some of my favorite songs with one of my favorite performers—baritone John Brancy, a young man of tremendous musical talent, blessed with a unique spark and a something I can only define as grace. I can't dance any more, but when I make music with John, I dance at the piano."



Howard and Ellen Poretsky with Pauli Overdorff (center)



Jeff and Beth (co-chair) Johnston



Susan Egert with the check-in crew

Photos courtesy of Diane Bondareff





## Save the Date!

**June 13**

Gathering in a private home on Gull Lake, Hickory Corners, Michigan

**July 30—August 1**

FSH Society 2010 International Patient and Researcher Network Meeting, Las Vegas, Nevada

**October 9**

Second Annual Walk 'N' Roll for FSH Muscular Dystrophy, Cape Cod, Massachusetts  
To register, go to [www.fshsociety.org/CapeWalk](http://www.fshsociety.org/CapeWalk)

**October 9**

Walk 'N' Roll, Southern California, Heritage Park, Irvine, California  
To register, go to [www.fshsociety.org/IrvineWalk](http://www.fshsociety.org/IrvineWalk)

Want more information about these events?  
Go to [www.fshsociety.org](http://www.fshsociety.org), or e-mail [info@fshsociety.org](mailto:info@fshsociety.org).



James Chin, Sr., member of the FSH Society Board of Directors, with wife Barbara and friends



Egert and Seslowe families



## The 2010 International Research Consortium & Research Planning Meeting is scheduled

The FSH Society International Research Consortium & Research Planning Meeting is set for October 21 & 22, 2010. The NIH-supported Boston Biomedical Research Institute Senator Paul D. Wellstone Muscular Dystrophy Cooperative Research Center for FSHD has kindly offered the use of its facilities for this meeting.

Silvère van der Maarel, Ph.D. (Leiden University), and Kathryn Wagner, M.D., Ph.D. (Kennedy Krieger Institute & Johns Hopkins School of Medicine), will be the scientific co-Chairs.

## FSHD AND MY JOURNEY OF FAITH

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FSHD has been a major consideration in every significant decision I have made in life. I chose a path to motherhood through adoption, out of concern for passing on this disease. And before we considered marriage, I asked the man who would become my husband to attend FSHD meetings and take time for careful reflection, to consider whether he felt prepared for a lifetime commitment to a spouse with significant physical limitations. Furthermore, I pursued education and work in the field of computer science, where I would rely on my mind rather than physical strength. So, it wasn't a surprise when FSHD became a factor in my decision to pursue a new path in life.

In addition to influencing many of my life's decisions, I believe FSHD has also greatly formed my character. It has taught me to cultivate an inner sense of grace, even though I might look a little shaky and less than graceful from the outside. It has taught me the importance of kindness and resilience, recognizing that we all have limitations and wounds in life, whether visible to the eye or not. FSHD has helped shape who I am, how I see myself in this world, and what is important to me. Most importantly, FSHD has given me opportunities to practice living with compassion and empathy, and to find meaning in life in spite of physical challenges and limitations.

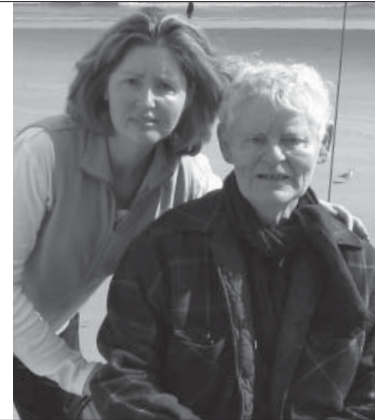
Being influenced in this way by FSHD created a growing feeling within me that I needed to pursue a more meaningful career while I was still able. FSHD added a sense of immediacy and purpose to my life, as I'm sure it does for many others facing illness or challenges. Although none of us can know how much time we have to pursue our hopes and dreams, having FSHD helped me realize the important things in life should not be put off until some future date. For me, that meant answering a call to ministry, and tending to others who are in need.

I have always been drawn to care for others; however, concerns over my increasing physical limitations and knowing I would be the one in need of care in the not-too-distant future led me to stay on a safer path. Last fall, however, I reached a point where I could no longer ignore the call to do something more, to make a greater difference to the lives of others. I decided to leave my job of nine years and a career of 25 years, and return to school to pursue ministry as a chaplain. This was one of the

most difficult decisions I have ever made, yet it has been one of the most rewarding. This type of decision might sound a little crazy for anyone, but for someone with a neuromuscular disease it tended to sound even crazier. I wondered if I was being foolish to leave the relative safety I had,

but it has prepared me to remain present and supportive to others facing similar loss and challenge. No two situations are ever the same, and even though I have my own experience of loss and limitation I would never claim to know how another person might feel in response to a loss of ability or

“FSHD has also taught me that life can be difficult and unpredictable, yet I can be filled with gratitude for the many blessings I have received, and continue to receive, throughout my life.”



to pursue a new career path filled with so many unknowns. I asked myself the same questions anyone would likely ask when making such a career change – would I be good at this? Would there be a place for me as a chaplain? Additionally, though, I asked myself whether I was physically up to such a change. I wondered whether I could be effective as a chaplain, even when my physical limitations became more apparent. Would a person in need find strength from a chaplain whose physical appearance was one of weakness rather than strength? I decided to take a leap of faith, knowing I would have too many regrets if I did not try my best to answer this call.

I knew FSHD had taught me important life lessons I could share with others; it wasn't until I returned to school that I began to consider the strengths FSHD has given me, that I now bring to my ministry as a chaplain. What I had considered as a potential weakness has become a strength.

FSHD has given me both the blessing and the burden of contemplating life's difficult realities at a very young age. Each of us has to face the difficult realities of our human condition; having FSHD simply encouraged me to think about these realities sooner rather than later, and decide how I might live my life in response to my particular human condition. This experience hasn't provided any easy answers or profound insight about how to accept loss,

even the same diagnosis. I do know that living as well and as gracefully as possible with FSHD has allowed me opportunities to practice making peace with the loss of some ability, while knowing there is still more loss to come. FSHD has also taught me that life can be difficult and unpredictable, yet I can be filled with gratitude for the many blessings I have received, and continue to receive, throughout my life. This is personal experience and hard earned wisdom that I bring with me as a caregiver, and I believe makes me a more empathetic and compassionate caregiver. Even without sharing my own challenges, I believe those I care for sense my calmness and comfort with life's hard realities.

I haven't often struggled with the difficult, and I believe unanswerable, “why” questions in life, such as “Why me?” or “Why FSHD?” And, I don't believe I have FSHD for a reason that is part of some greater plan. Instead, my faith challenges me to consider what I will do with the knowledge and experience FSHD has given me; it asks me to trust that I can do something good with this experience. When viewed from this perspective, FSHD has given me a sense of purpose in life and it has prepared me to become a better caregiver.

My father died unexpectedly, from respiratory complications due to FSHD, just

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## The Roaring 20's: A Night of Wonderful Nonsense

The Culinary Institute of America, Hyde Park, New York, together with Society members Gary and Arlene Cohen, hosted a benefit dinner for the Society on January 30, 2010.



Gary Cohen and Justin Cohen



Arlene and Gary Cohen and friends

### FSHD AND MY JOURNEY OF FAITH

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a few weeks after I made my decision to pursue chaplaincy. This tremendous and sudden loss reinforced to me that life is too short to put off until later those things that are important to each of us. I continue to be moved when I hear the inspiring stories of so many individuals with FSHD who live life so fully. I am reminded that those of us with FSHD are a resilient and resourceful community of individuals who have so much to offer, to ourselves and to others. If I am able to bring hope and comfort to others through my ministry, then I will reach my own goals of living life fully and making a caring difference. Additionally, I remember that through the kindness and support of others, especially Dan, Nancy, and Jenny at the FSH Society, Elicia Estrella at Children's Hospital Boston, and Regina Brock-Simmons and Dr. Wagner at Johns Hopkins University, we will do great things in response to the challenge of FSH muscular dystrophy.

#### ► Have you made a gift to the Society in 2010?

The FSH Society is a world leader in combating muscular dystrophy. It has provided over \$2 million in seed grants to pioneering research worldwide and has created an international collaborative network of patients and researchers.

If you are not already a member, won't you join in this effort? Please return your membership gift, or another gift, in the enclosed envelope. Or contribute online at [www.fshsociety.org](http://www.fshsociety.org). Go to Contribute, and select the gift category you wish to make. Thank you.

#### ► Volunteers are still needed for muscle biopsy study

For information, go to [www.fshsociety.org](http://www.fshsociety.org), or contact Jenny Lazzaro at the FSH Society, (617) 658-7877 or [jennifer.lazzaro@fshsociety.org](mailto:jennifer.lazzaro@fshsociety.org), or Genila Bibat, M.D., at Johns Hopkins, (443) 923-2778.

#### ► Is your e-mail address current at the Society?

If we do not have your current e-mail address, and if you want to be sure of receiving up-to-the-minute information from the Society as news breaks, please send your e-mail address to us at [jennifer.lazzaro@fshsociety.org](mailto:jennifer.lazzaro@fshsociety.org). Thank you.



#### ► Do you follow the Society's Facebook page?

Since its launch in early 2008, the Society's Facebook page has attracted hundreds of viewers and many fans. Go to [www.facebook.com](http://www.facebook.com) and sign up. It is free and easy. Search on FSH Society and join the discussion. Bookmark it and come back often.

It is the editorial policy to report on developments regarding FacioScapuloHumeral Muscular Dystrophy (FSHD), but not to endorse any of the drugs or treatments discussed. We urge you to consult with your own physician about the procedures mentioned.

## End of Tax Season Celebration for FSH Muscular Dystrophy raises \$23,000 for Society

Christopher and Ellen Stenmon hosted the 12th annual End of Tax Season Celebration, a very successful fundraiser, on April 30, at Florian Hall in Dorchester, Massachusetts. Guests enjoyed good food and drink, music, dancing, a door prize, a silent auction and a raffle. They had great fun and supported a great cause. Chris is a member of the Society's Board of Directors.



Chris and Ellen Stenmon (left) and Stenmon family



Kevin Pellerin and other dancers



Judge Ed Boyle introduces Daniel Paul Perez, President and CEO, FSH Society, Inc.



Friends and family



Friends having a nice evening together



Happy party-goers