



FSH Watch

A publication of the Facioscapulohumeral Muscular Dystrophy Society
Connecting the community of patients, families, clinicians and investigators

On the road to treatment: Biomarkers will lead the way

By Kathryn R. Wagner, M.D., Ph.D., Director, Center for Genetic Muscle Disorders,
 The Kennedy Krieger Institute, and Associate Professor of Neurology, Johns Hopkins
 School of Medicine, Baltimore, Maryland

At meetings such as the biennial FSH Society International Patient/Researcher conference, scientists working on FSHD frequently voice their discouragement with the lack of “biomaterials” to study. Essentially, we do not have sufficient blood, cells, and tissue from FSHD patients in order to answer important questions, such as “what genes are over- or under-expressed in FSHD muscle leading to weakness,” and “do FSHD muscle cells behave differently than normal muscle cells.” This barrier in FSHD research is now being eliminated thanks to the FSH Society, the U.S. National Institutes of Health and families affected with FSHD.

One of the key components of the year-old Senator Paul D.

Wellstone Muscular Dystrophy Cooperative Research Center for FSHD Research, established in 2008 and based at the Boston Biomedical Research Institute (BBRI), is a “Cell Core” which is open to researchers throughout the world. The Cell Core is a repository of biomaterial from FSHD patients and their first degree family members (siblings, parents or adult children). An important and unique feature of the Cell Core program is that muscle cells obtained from volunteers are “immortalized” so that they will reproduce in a laboratory dish nearly indefinitely and not be exhausted. Biomaterials contributed to the Cell Core will therefore advance not just the research of the investigators within the FSHD Wellstone Center but

that of many other scientists now and in the future.

One of the questions that Louis Kunkel, Ph.D., Harvard Medical School and Children’s Hospital Boston, and I are most interested in addressing through this program is whether we can identify a specific “molecular signature” of FSHD disease. We anticipate that an FSHD

“molecular signature” might be a half dozen genes that are turned on and a few that are turned off in FSHD, compared to normal muscle. The Cell Core program has been uniquely constructed to allow us to answer this question. We are obtaining muscle from the biceps, which is involved early in the disease, and from the deltoid, which is involved later, if at all. We can then

study the gene expression in these two muscles of the same individual and in the muscles of their unaffected family members to define the molecular signature that spells FSHD.

Another term for this FSHD signature is “biomarker.” A biomarker is “a characteristic that is objectively measured and evaluated as an indicator of normal biologic processes, pathogenic processes, or pharmacologic response to a therapeutic intervention” (*Clinical Pharmacology Therapeutics* 200; 69: 89-95). One of the reasons that it is so crucial to identify a biomarker of FSHD disease is that it facilitates drug development. Diseases that have good biomarkers have many more drugs developed to treat them. For example,



Mary Mruk and daughter, Ashley Mruk, participated in Dr. Wagner’s research study in summer 2009. Ashley is the youngest participant to date, at age 18.



Thank you, Mrs. McKenney & kindergartners!

In Spring 2009, Mrs. McKenney’s kindergarten class at Hopewell School, Taunton, Massachusetts, made an ABC quilt as a class project. Each child made one of the alphabet letter blocks. They sold raffle tickets to others in the school, raising nearly \$300 for FSHD research. Mrs. McKenney’s friend has a child with FSHD and this is the second year she has led this class project. A first grader won the quilt.

diabetes has the biomarker glycosylated hemoglobin, which not only helps define the disease but also indicates the degree to which blood sugars are under control in this disease. Diseases such as Amyotrophic Lateral Sclerosis (ALS) and Alzheimer’s disease do not have good biomarkers and have very few effective therapeutics. There is not a currently defined biomarker of FSHD and, as this audience is painfully aware, there has not been a single drug developed to combat the disease.

If we can define a molecular signature or biomarker of FSHD then we can begin to ask, which therapies move this signature towards a signature of normal muscle. For example, if the FSHD biceps has more expression of genes x, y, and z but less expression of genes a, b and c than FSHD deltoid and normal muscle, then we would investigate drugs which

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Letter from the Executive Director

Dear Friends,

In recent months, many of you have asked me “how is the Society doing”? In other years, you would likely be curious about new research findings, new lines of inquiry and other indicators of progress. But now your question is more often out of your concern for the financial health of the Society.



Nancy Van Zant

We are happy to share with you that, as of this writing, the FSH Society remains strong financially. In 2009, we have made new research awards and renewed awards where we have had continuing commitments totaling \$210,000, with an additional \$174,000 in grants awaiting approval of the board of directors at the end of October.

All that we accomplish is dependent on philanthropy—membership and other gifts—from you. To date in 2009, the Society is “holding its own” in contributions compared to the same time in 2008.

Like you, we are eager to have funds available to respond to investigators when they approach the Scientific Advisory Board requesting support for new lines of research. I hope you will give generously in the coming months and join with others to keep the FSH Society strong. Progress remains foremost on our minds.

All good wishes to you, and thank you.

Sincerely,

Nancy Van Zant
 Executive Director

Charitable IRA rollover opportunity

In October 2008, the U.S. Congress approved and the President signed legislation that included the immediate and retroactive extension of the popular Pension Protection Act of 2006 provision which allows U.S. taxpayers over 70½ to make tax-free distributions from their traditional and Roth IRAs (Individual Retirement Accounts) directly to charity in 2008 and 2009.

Check with your advisors about the best ways to take advantage of this opportunity to give to the FSH Society in 2009. As always, the FSH Society will also be pleased to assist you in any way possible.

Correction: Summer Watch

The hard copy of the FSH Watch Summer 2009 Research newsletter contained some errors.

In the article by Richard Lemmers, Ph.D., at page 11, in the second full paragraph, the first three sentences should read as follows:

“With the establishment of the Fields Center for FSHD and Neuromuscular Research

www.urmc.rochester.edu/fields-center/ we have now created in collaboration with Dr. Rabi Tawil (University of Rochester Medical Center) standardized clinical and genetic assessment protocols.

These protocols are now fully implemented in the Fields Center and we hope these protocols will prevent future mistakes in DNA diagnosis. With the Fields Center we are also planning for a best practice meeting for DNA diagnosis in FSHD early next year.”

In the article by Yvonne Krom, PhD, at page 13, in the third paragraph, the seventh sentence should read as follows:

“In fact, this collaboration is now further consolidated in the Fields Center for FSHD and Neuromuscular Research at the University of Rochester Medical Center headed by Professor Rabi Tawil.”

FSH Society advocacy efforts in Washington, D.C., continue to yield big gains in research funding

U.S NIH funds \$2 million for “FSHD iPS Cells: Modeling Disease Mechanisms, Genetic Correction and Cell Therapy”

By Michael Kyba, Ph.D., University of Minnesota, and Daniel Paul Perez, FSH Society, Inc.

A recent breakthrough in stem cell science—applied to FSHD—is the target of a study by four experts led by principal investigator Michael Kyba, Ph.D., of the University of Minnesota. The team, which also includes Minnesota scientists Rita Perlingeiro, Ph.D., and John Day, M.D., and Baylor College of Medicine’s Thomas Zwaka, M.D., Ph.D., received nearly \$2

million in funding from the National Institute of Arthritis and Musculoskeletal and Skin Diseases. While it is known *what* variation in the DNA is associated with FSHD, it is not known *how* it causes the progressive muscle loss. By removing cells from individuals with FSHD and re-programming them to become induced pluripotent or iPS cells similar to embryonic stem cells, the team expects to gain new insights into the genetic basis of the disease and discover

possible new cell therapies.

Work in Dr. Kyba’s laboratory has led to a new theory for FSHD that postulates interference with the molecular mechanisms governing regeneration of muscle by a gene encoded by the D4Z4 repeats, known as DUX4. To test this theory, as well as to study the regulation of D4Z4 in skeletal muscle regeneration, large quantities of skeletal muscle stem cells are required, preferably obtained before the muscle is affected with disease. Dr. Perlingeiro’s laboratory has recently developed a method of producing just such cells from embryonic stem cells, and the team is applying this method to FSHD-bearing iPS cells.

Dr. Day, who has made groundbreaking advances in the understanding of myotonic dystrophy, is the director of the Paul and Sheila Wellstone Muscular Dystrophy Center at the University, and is providing clinical expertise. Dr. Zwaka is a leader in the genetic modification of human ES cells, and will be testing strategies to modify chromosome 4 in these cells in such a way that the FSHD-causing mutation is eliminated. The long-term objective of the team is to enable a cell therapy from genetically corrected iPS cells.

The Minnesota team is currently recruiting both postdoctoral scientists to participate in the research, and individuals with FSHD, particularly those rare individuals without a contraction on chromosome 4, to provide cells for ongoing studies. Dr. Kyba can be reached at halle071@umn.edu.

The work that laid the foundation of this grant was generously funded by the Pacific Northwest Friends of FSH Research, and by the Dr. Bob and Jean Smith Foundation. The work was also supported by the Fields Center for FSHD and Neuromuscular Research, which provided patient material. In addition, work in Dr. Kyba’s laboratory on DUX4 has been supported by the FSH Society through a Marjorie Bronfman fellowship

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Judith Seslowe, M.A., elected to Society’s Board of Directors

Judith Seslowe joined the Board of Directors in June 2009, after serving the Society for many years in her suburban New York community. She holds undergraduate degrees from the University of Pennsylvania and City College of New York, as well as an M.A. from Columbia University Teachers College. Many homes in Westchester County have beautiful gardens and lawns, thanks to Judith Seslowe Landscape Design. Judy founded this company and has worked in residential landscape for many years.

She and her husband Kenneth



Judith Seslowe, M.A.

have two married children, Emily and Jonathan, ages 40, and 38, respectively, and four grandchildren ranging in age from four to 13. In addition to her family and work, Judy enjoys bridge, travel, music, theater and art.

Judy has raised significant funds for the FSH Society through her personal giving and by chairing the concert in New York for the Society’s benefit in 2009. Judy is optimistic about the prospect of treatments and wants to continue to help advance progress through her work in New York. She is also a resource and peer to other individuals with FSHD.

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.

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Biomarkers,

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decreased expression of x, y and z and/or increased expression of a, b, and c in muscle. In parallel efforts, we have been advocating to pharmaceutical companies in the process of developing drugs for muscle disorders, that they obtain muscle samples and define their drugs' gene expression profile so that we can begin to determine the appropriateness of these drugs in the treatment of FSHD.

True to form, the FSHD community has already shown its dedication and generosity in accomplishing the goals of the biomarker program and the larger FSHD Wellstone Cell Core program. Eleven families have participated or plan to participate in contributing muscle and blood samples. These families have come from all over the U.S., and the FSH Society has helped to pay for their travel to Baltimore, where I have enjoyed meeting them. Visits have been for three to four days during which we screen the family members to make sure that their participation is both helpful and safe. A history and physical exam are performed, blood is obtained and muscle is biopsied from the deltoid and biceps. Frozen muscle is immediately sent to numerous Wellstone MD CRC investigators for gene, protein and tissue studies, and small pieces of muscle are kept alive in special broth and sent to BBRI to begin the process of isolating and immortalizing cells.

Although it may not seem like a favorite activity for a family reunion or summer vacation, the response of participants (see column, right) has been overwhelmingly positive. I think there are a number of reasons for this. First, the FSH Society, researchers and staff all recognize the extraordinary contribution that volunteers are making and treat them accordingly. Second, shared challenges, be they basic training in the military, medical internship or muscle biopsy, create camaraderie among people. Third, and most importantly, as the FSHD community has seen from its participation in clinical trials, being able to contribute to the solution, to take control of some aspect of this disease, is empowering.

Our trip to Johns Hopkins Hospital

By Mary Rieger

On a Wednesday in August, my mother and I headed to Baltimore to participate in a research study being conducted at Johns Hopkins Hospital.

As readers know, the principal investigator is Kathryn R. Wagner, M.D., Ph.D., and the research coordinator is Regina Brock-Simmons. The research being carried out is to collect and store blood (DNA) and muscle tissue from people affected with FSHD and a first degree relative who is unaffected by FSHD.

Future research with these specimens should show which genes and proteins are different in FSHD

muscle compared to normal muscle, should help understand the cause of the disease, how the disease changes over time and why some muscles are affected more than others.

On Wednesday we had pre-operational procedures including all of the basics, physical examination, testing of muscle strength and lots of blood. They took eight containers of blood out of us! We did manage to survive though, and the mosquitoes did not want anything to do with us that night!

On Thursday we returned to the outpatient center for surgery. We were placed in rooms next to each other in the prep area and were able to wander back and forth visiting each other in our hospital gowns. The hospital staff at

Johns Hopkins was wonderful, including Dr. Wagner and Regina. The nurses were all so friendly and talkative and kept us laughing even though we were a little nervous. They all made the visit very unforgettable.

Mom went into surgery for her muscle biopsy at 1:30 p.m. and I followed at 2:30. We were then in post-operation together, eating our snacks and having fluids. Oh and extra strength Tylenol! We were released by 3:30 and headed back to the hotel to relax. On Friday, we left Baltimore and headed to Cape May, New Jersey, for a little R & R with our new matching bandages. Talk about strange looks! It is amazing how many people

go up to complete strangers to ask what happened. We were always very happy to share our story of our matching wounds!

I had a wonderful time throughout the whole trip! I am so very blessed to have my Mom! She did not even hesitate when I asked her to participate with me in the study. This was our first "vacation" ever where it was just my mom and me, and what a great, memorable time it was. She truly would do anything for me and has proved this over and over again throughout the years. To know that I have her by my side to help me fight this battle and hopefully find a cure, I am not scared.

Thanks Mom, I Love You!



Mary Rieger, right, and her mother, Autumn Hammond

Volunteers are still needed for muscle biopsy study.

Go to www.fshsociety.org for more information, or contact Regina Brock-Simmons at Johns Hopkins, (410) 502-7220, or Jenny Lazzaro at the FSH Society, (617) 658-7877 or jennifer.lazzaro@fshsociety.org

Advocacy efforts yield big gains,

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grant to Darko Bosnakovski, Ph.D., a postdoctoral fellow in Dr. Kyba's group.

Dr. Kyba adds that he would like the readers to know that there are additional important ways that the FSH Society and Daniel Paul Perez, its CEO, have helped to make these opportunities possible including supporting research networking meetings and annual symposia and, perhaps most importantly, through advocacy work in Washington, D.C., and with the NIH. Dr. Kyba said, "We thank the FSH Society, and we thank you for your constant and continued support."

The FSH Society Future Fund – It's about time and money

By Amy Bekier

Past... present... future. They're all intertwined, aren't they? Those who have left us legacies from the past are helping to pave our way into the present. Those who intend to leave us a legacy today are paving it forward into the future. This topic about "Time" leads me to talk to you about the absolute importance of the FSH Society Future Fund.

The FSH Society is our most significant advocate. Who stands up to make sure we are counted when it comes to legislation and government funding? Who is there to hold our and our family's hands when a personal crisis arises? And, who organizes the patient conferences that enable us to connect with up-to-the-minute information and with friends who are going through similar experiences? Whose Scientific Advisory Board awards grants so that scientists can search worldwide to find a specific treatment and cure for FSHD? The FSH Society does all of this and more. They try diligently to educate the world about the fact that this devastating disease is the most prevalent of muscular dystrophies yet it remains underrepresented and underreported. The Society's activism in pursuing treatment has spurred other organizations to follow its lead.

This brings me to the topic of "Money." Without money the wheels of progress and the seeds of hope come to a screeching halt. *This is the reason that I have chosen to join the FSH Society*

Future Fund by including the Society in my will. Not only is there great logic in joining the fund as well as possible tax advantages, but it feels wonderful and prestigious to know that I will be able to make an impact in destroying my family's lifelong disease, perhaps many years beyond my own life. One need not be wealthy to leave a legacy, but I must admit that there is a little immortality and ego that goes along with the bequest.

There are so many different ways to join the Fund. It can be done simply by listing the FSH Society in one's will. A knowledgeable attorney can set up a Charitable Remainder Annuity Trust for those concerned that loved ones may need continuing financial support. In some cases, IRA money might receive possible tax advantages. Of course all of these and other possibilities should be pursued with a tax and estate advisor. And make sure to let the FSH Society know your intentions.

Even if a treatment or cure were found tomorrow, money would still be needed for testing, distribution, patient care and resources. I pray that someone's past legacy will help find a cure for me today. And I pray that my legacy will help someone in the future. Past... Present... and Future are all intertwined. The FSH Society's Future Fund ...it's all about Time and Money. Please consider it. And above all else, please make sure to join the FSH Society or renew your membership! *Amy Bekier lives in San Diego, California. She became an artist, www.amyzoart.com, after she retired as an insurance and investment agent. Amy has FSHD and is an FSH Society and Future Fund member.*

Respiratory Insufficiency Can Occur in FSHD

Be Aware of Your Respiratory Status

► Evaluation of the symptoms and signs of respiratory insufficiency should be sought during routine clinic visits in patients with moderate to severe FSHD.

► In standard practice, trauma (ER, ICU), surgery and anesthesiology settings, care should be taken not to suppress respiratory drive with narcotics unless it is a situation of palliative care.

► Oxygen supplementation can be detrimental to patients with hypercarbic (high CO₂) respiratory failure.

► Oxygen should generally not be administered to patients with hypercarbic respiratory insufficiency unless BiPAP or similar ventilatory support is also being used.

► Your physician and a pulmonologist can help you periodically monitor CO₂ levels in the office or pulmonary function lab in the hospital.

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For more information visit:
www.fshsociety.org

Following an article about "Anesthetic Concerns," in the Winter 2008 Watch (pages 8-9), the Society offered to prepare a fact sheet to be carried by patients and their families. Since then, we have reviewed this topic with physicians and patients, and have determined that the issues involved are so specific on a case-by-case basis that it is not in the best interest of people with FSHD to offer a generic set of guidelines. However, we are able to provide a cutout (above) that may facilitate a dialogue between you and your physicians, including a pulmonologist and anesthesiologist.

Chronic pain in people with FSHD

By Gregory T. Carter, M.D., M.S., Professor of Rehabilitation Medicine, University of Washington School of Medicine, Seattle, Washington

Why study pain in FSHD?

Recent research suggests that chronic pain may be a significant problem in many people with chronic neuromuscular disease (NMD), including all forms of muscular dystrophy (MD). The most common form of MD is facioscapulohumeral muscular dystrophy (FSHD), and this has been the focus of our recent studies. A study of four individuals with FSHD who identified pain as their most disabling symptom and made three to seven separate pain complaints has been reported by Bushby, *et al.* In addition, Abresch, *et al.*, found that 83% of a sample of 811 individuals with various NMDs, including 64 individuals with FSHD, reported at least some ongoing pain problems. Moreover, the frequency and severity of pain in their sample of patients with FSHD was significantly greater than levels of pain reported by the general U.S. population.

Our group initially surveyed individuals with a variety of NMDs,

including patients with FSHD, and found that 73% of the sample as a whole, and 89% of patients with FSHD, reported pain problems, with 27% of the overall sample reporting severe pain (19% of patients with FSHD). Pain was reported to interfere moderately with a number of activities of daily living across all of the NMD diagnostic groups (range of interference ratings, 2.6 to 4.63 on 0 to 10 interference ratings scales) and to occur all over the body (least common, abdomen/pelvis at 16%; most common, back at 49%). Medications were the most common pain treatment used by these patients, with ibuprofen, aspirin, acetaminophen, opioids, neurontin, and muscle relaxants being used by 50% or more of the patients with pain.

Although the preliminary findings from our group and others indicated that chronic pain can be a serious problem for many with FSHD, much remains unknown about the nature and scope of pain in these patient populations. Most of the research on pain that has been performed with patients with FSHD has reported findings from a mixed population of patients with limited sample sizes for particular diagnoses. This limits both the reliability and generality of the available findings. Descriptive analyses regarding pain with larger samples of patients with specific diagnoses would provide greater reliability of the findings, and would allow us to confirm, or to question, previously published data concerning pain in patients with these conditions.

How we investigated pain in FSHD

More recently we recruited FSHD participants from the National Registry of Myotonic Dystrophy and Facioscapulohumeral Muscular Dystrophy Patients and Family Members (<http://www.dystrophyregistry.org>) as well as our own University of Washington Muscular Dystrophy Association (MDAUSA) Clinic. Our survey included questions asking for demographic information, NMD-related information, pain intensity, pain interference, pain

location, and pain treatments. All participants provided basic demographic information about their gender, age, race/ethnicity, educational level, marital and employment status. They also provided information about their diagnosis, including approximate date of diagnosis, type of physician who made the diagnosis, whether or not they had received a DNA confirmation of diagnosis, and the extent of their mobility limitations and use of assistive devices. Average pain intensity over the past week was assessed using a 10-point numerical rating scale (0, “no pain” to 10, “pain as bad as could be”) taken from the *Grading of Chronic Pain* scale (GCP). Pain interference with daily activities was assessed using a twelve (12) item interference scale adapted from the *Brief Pain Inventory Pain Interference* scale (BPI). Participants were asked to rate the extent to which pain interferes with twelve (12) specific activities during the preceding week on a 0 (“does not interfere”) to 10 (“completely interferes”) scale.

The original BPI Pain Interference scale includes seven items: general activity, mood, walking ability, normal work (including both work outside the home and housework), relations with other people, sleep, and enjoyment of life. We modified the original scale to make it more valid for persons with FSHD by changing the “walking ability” item to read “mobility (ability to get around),” because many people with FSHD may be unable to walk, even when pain-free. We also added five items: interference with self-care, recreational activities, social activities, communication with others, and learning new information or skills, to obtain a broader-based sample of interference domains that both are particularly important to persons with physical disabilities and could potentially be impacted by pain. The original BPI Pain Interference scale has data supporting its reliability and validity as a measure of pain interference in persons with cancer. Our modified version of this scale has demonstrated high levels of

Save the date

For gatherings of FSH Society members and friends!

- ▶ Birmingham, Alabama
Afternoon gathering in a private home, Sunday, October 25
- ▶ Hamilton, New Jersey
Luncheon, Sunday, November 1
- ▶ Las Vegas, Nevada
July 30—August 1, 2010
FSH Society International Patient and Researcher Network Meeting

For more information, contact the FSH Society office,
info@fshsociety.org
or (617) 658-7878

Chronic pain in people with FSHD

internal consistency and validity through strong association with pain intensity in samples of persons with various forms of chronic disability.

Participants were asked to indicate whether or not they experience bothersome pain in one or more of seventeen (17) specific body sites (head, neck, shoulders, upper back, lower back, arms, elbows, wrists, hands, buttocks, hips, chest, abdomen/pelvis, legs, knees, ankles and feet). If they did report pain in a specific location, they were asked to give the approximate date that pain problem began and to rate average pain intensity in that location over the past week on a 0 (“no pain”) to 10 (“pain as bad as could be”) scale. Participants were asked to indicate if they were currently using or had ever used any of twenty-five (25) specific pain treatments (physical therapy, nerve blocks, biofeedback/relaxation training, acupuncture, magnets, massage, hypnosis, counseling/psychotherapy, mexiletine, neurontin, tricyclic antidepressants, narcotics/opioids, acetaminophen, aspirin/ibuprofen, valium, tegretol, baclofen, TENS units, anticonvulsants, chiropractic adjustments, heat, ice, cannabis (marijuana), strengthening exercises or range of motion exercises). They were also asked to indicate the amount of relief that each treatment they had tried provided on a 10-point scale (0 being no relief to 10 being “complete relief”).

What did we find out about pain in FSHD?

The vast majority of individuals with FSHD (82%) reported experiencing pain, with the average pain intensity being in the moderate range overall (4.45). Pain interfered the most with recreational activities and mobility or ability to get around. Despite this, FSHD participants are more likely to be working full time than what has been reported in other forms of NMD, with 33.9% of FSHD participants in our survey working full-time. Pain interference with each activity was also moderately to strongly

associated with pain intensity. The most frequent pain sites for FSHD patients were the low back (74%) and the legs (72%). Physical therapy (50% of all FSHD respondents) was the treatment most often tried for pain management, with application of heat being the second most common. Over the counter pain medications and exercise follow close behind with ibuprofen (32% patients as a whole; 42% FSHD group), acetaminophen (36% patients as a whole; 40% FSHD group), and strengthening exercise (35% patients as a whole; 40% FSHD group). The treatments that provided the greatest pain relief were not necessarily those that were most frequently tried or still used. The highest average relief rating (8.29 on a 0 to 10 scale) came from severe pain patients reporting using cannabis (marijuana) for pain relief. However, only 7% of the entire sample, and 12% of those with severe pain, reported ever trying cannabis for pain relief. Other pain treatments that were reported as relatively more effective included opioids (6.49 average relief for all patients, 6.70 average relief for patients reporting severe pain), nerve blocks (5.38 average relief for all patients, 5.33 for those with severe pain), massage (5.16 average relief for all patients, 6.05 average relief for patients reporting severe pain), and chiropractic manipulation (5.32 average relief for all patients, 5.53 average relief for patients reporting severe pain). Less than 30% of all participants reported trying any of these other methods of pain relief (only 8% had received nerve blocks). Most of the other treatments, including hypnosis (2.00 relief for all patients, 2.00 for severe pain patients), magnets (2.50 for all patients, 1.14 for severe pain patients), and counseling (3.00 for all patients, 2.78 for severe pain patients) were rarely tried.

Pain severity correlated with severity of disability, with more physically disabled people with FSHD reporting the most difficulty with pain. In another study just completed, we were able to show that higher levels of perceived social support were associated with less pain interference and better psychological

functioning in people with FSHD, even after controlling for pain intensity and demographic variables. These findings are generally consistent with other research of people with disabilities and pain as well as with studies of people with pain and other health problems. The consistent associations found in this body of research raise the possibility that social support, or at least one’s perceived access to support, may act as a buffer for maintaining physical health, and psychological functioning, in persons with disabilities and pain.

Padua, *et al.*, just published a study assessing Quality of Life (QoL) and evaluating the occurrence and characteristics of pain in FSHD patients. They did a prospective study using several different assessment tools, including: clinical, genetic, QoL, pain, and depression. In the 65 patients they enrolled, QoL was significantly reduced in FSHD patients, mainly in physical domains. Their study in Italians with FSHD confirmed what we saw here in the United States: pain is a commonly occurring, significant problem in FSHD patients. In their study, more than half of the patients complained of at least moderate pain. Women complained of slightly higher levels of deterioration in the emotional aspects of QoL than men. As we had also noted, physical disability correlated with more severe QoL deterioration. Their study confirmed our work but also noted some gender differences: that women may suffer greater deterioration in the emotional aspects of QoL compared to men with FSHD.

What can we conclude from this?

Several studies now indicate that pain is common in patients with FSHD and can significantly impair quality of life. This underscores the need to identify and provide effective pain treatments for patients with FSHD. Indeed, pain interfered not only with many basic activities including communication with others, relations with other people,

Chronic pain in people with FSHD

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learning new skills, and self-care, but also with activities that require the use of muscles, e.g., mobility, normal work, and recreational activities. Overall, the sites of pain reported by these patients reflect the body areas that are commonly affected by these muscular dystrophies, e.g., low back and legs as most common, chest, buttocks, and head as relatively less common. There may be some gender

differences as well, with women suffering greater deterioration in the emotional aspects of quality of life.

Clearly what is needed is more research! We really need to look at different strategies to treat pain in FSHD. This includes pharmaceutical agents as well as physical medicine and rehabilitation techniques, including psychosocial and vocational domains.

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Gifts of stock to the FSH Society

As you review your financial affairs as 2009 draws to a close, it will be important for you to consider whether investment market conditions make it advantageous for you to make a gift of stock to the FSH Society at this time.

Consider the tax benefits

A gift of appreciated securities held for more than one year may provide significant benefits to you as a contributor, such as:

Providing you a charitable income tax deduction for the fair market value of the gifted securities as of the date of gift

Eliminating capital gains tax that would ordinarily become due if you had sold the appreciated securities on the open market and donated the proceeds from the sale to charity

Claiming your charitable deduction against up to 30% of your adjusted gross income. Any unused deductions can be carried forward over the next five years

Providing a way to help you to achieve your long-term financial objective of reducing your income and estate taxes.

Caution!

Tax benefits are lost if:

- ▶ the stock is sold and then the proceeds are given
- ▶ the stock is worth less than you paid for it
- ▶ the stock has been held for one year or less.

For more information, contact the FSH Society at (617) 658-7878, or go to the website, www.fshsociety.org and click on Contribute and then select Gifts of Stock. You should also consult with your financial advisor before initiating a charitable gift of stock.

International Research Consortium to meet in Watertown, Massachusetts, November 2009

The annual FSH Society International Research Consortium (IRC) Workshop is scheduled for Monday, November 9, 2009, at the Boston Biomedical Research Institute (BBRI), Watertown, Massachusetts.

Kathryn R. Wagner, M.D., Ph.D., Director, Center for Genetic Muscle Disorders, The Kennedy Krieger Institute, and Associate Professor of Neurology, Johns Hopkins School of Medicine, Baltimore, Maryland, and Professor Silvère van der Maarel, Ph.D., Leiden University Medical Center, Department of Human Genetics, and Co-director Fields FSHD Center, will be the clinical and research Co-chairs for this meeting. Daniel Paul Perez, FSH Society President and CEO, is organizing the meeting.

This FSHD International Consortium Workshop is the only annual platform for clinicians and basic scientists to be informed about the latest developments in FSHD. For many years now, this workshop has provided the FSHD community a forum to present and

discuss new developments, reinforce collaboration and facilitate new initiatives. The FSHD IRC convenes annually to pursue three goals:

1. to discuss the genetic and biological mechanisms and patho-physiological causes of FSHD,
2. to improve diagnostic techniques and criteria for FSHD, and
3. to consider and evaluate new and existing therapies for the disorder.

The presence of these investigators and the platform presentations is validating evidence to the community of rapid developments on numerous fronts in FSHD. We are seeing increasing momentum in basic and clinical research initiatives as well as the development of promising, potential treatments for FSHD and other muscular dystrophies. Additionally, there is an increase in government, non-profit, and private funding and in international collaboration of volunteer health agencies and FSHD patients.

Research planning day to follow Research Consortium

In response to requests from scientists in the FSHD research community, the Scientific Advisory Board of the FSH Society will once again lead a research planning process, the first since July 2006. Researchers from the Consortium Workshop on November 9 are invited to participate as well as representatives from MDAUSA, AFM, and the NIH program staff. They will convene to look at recent investigations and to plot a course of activity that best addresses next steps in FSHD research. The FSH Society will use these findings to help plan our near term investments in research. The proceedings will become a plan for FSHD research over the next 24-36 months. The FSH Society will publish the compendium of the presentations, in print and online, as a benchmark in the research program directions for FSHD.

The group will meet on Tuesday, November 10, 2009, at BBRI. Experts in the fields of muscle disease, muscular dystrophy, neuromuscular disease and molecular genetics will help draft the compendium and research plan.

Society members gather in June and July

Members and friends of the FSH Society had a picnic and pool party on a chilly Saturday afternoon in Alexandria, Minnesota.



Thanks to the Wagners for hosting a great day.



Members of the FSH Society meeting and greeting near the Twin Cities in Minnesota



Thanks to friends around the Twin Cities for organizing and sharing a picnic lunch with all of us.



Thank you to the Levinson and Williams families for hosting a great afternoon in Denver, Colorado, and to many FSH Society members and friends for joining us!

Are you a member of the FSH Society?

Have you made a gift to the Society in 2009?

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. Go to **Contribute**, and select the gift category you wish to make. Thank you.

Patients & families to gather in Las Vegas, July 30 to August 1, 2010

The 2010 International Patient/Researcher Network Meeting will be held at the Paris and Bally's Hotels in Las Vegas, July 30-August 1. These hotels have been selected for their accessibility, convenient location, and good value. They are a part of the same complex and share meeting space.

The conference will begin with registration and lunch at noon on Friday, July 30, and conclude after lunch on Sunday, August 1.

Lectures and small group discussions are planned to bring the most current advances in FSHD research, including the new NIH Senator Paul D. Wellstone Muscular Dystrophy Cooperative Research Center for FSHD Research, to the FSHD community of patients, families and scientists. There will be both formal and informal forums to share experiences of living with FSHD, practical and clinical information for daily living such as



breathing and respiration, nutrition, travel and leisure, exercise and physical therapy, patient advocacy, and other topics you have requested. It is also possible that a keynote speaker will discuss new classes of drugs to improve muscle health.

We will also provide opportunities for different age and care groups to gather—teenagers and young adults, parents, siblings, IFSHD families, if you are all present.

The program and the registration fee are developing. We expect to offer one registration fee that covers three lunches; an optional festive dinner may be available for Friday night, likely at a venue outside the hotels. More details will be made available on the Society's website as planning proceeds, and we will also mail registration materials to you.

For Bally's or Paris hotel reservations,

you may call 1-877-603-4389. The hotels are holding a block of rooms at \$89.00 (Bally's) per night (single or double occupancy) and \$120.00 (Paris) per night (single or double occupancy) plus taxes. There is no charge for parking. The **group code** for our conference rate for Bally's is **SBIPRO** and for Paris, **SPIPRO**. These facilities have many wheelchair accessible guestrooms, including a total of 100 rooms with roll-in showers. For the best selection of accessible rooms, please make your reservations early. The closing date for the Society's block of rooms is July 9, 2010. Here is how to make your hotel reservation online:

For Paris

<http://www.harrahs.com/CheckGroupAvailability.do?propCode=BLV&groupCode=SBIPRO>

For Bally's

<https://www.harrahs.com/CheckGroupAvailability.do?propCode=PLV&groupCode=SPIPRO> for Paris.

We are here to help! Society launches Peer-to-Peer Team

Many readers recall a time when they had a new diagnosis of FSHD and needed someone to speak with at once. Others living with FSHD want a knowledgeable and compassionate person for a conversation from time to time. The Society has formed a Peer-to-Peer Team of individuals with many experiences—as patients, parents, and other family members who may share some of your experiences.

FSH Society volunteers are available to answer your questions and help you get through a difficult time. Our goal is to respond within 24 hours.

Whether you have a new diagnosis of FSHD or have been living with FSHD for many years, we understand that you may experience a range of reactions, including anger, sadness, fear of the future, and feeling isolated with the disease. Or, you may be the parent of a child with FSHD or Infantile FSHD and wish to speak with

another parent. Whatever your feelings, they are normal and you are not alone.

Life with FSHD poses challenges for every member of the family. Whether you are affected with FSHD, the parent of an affected child, or the loved one of a person affected, it takes time to adapt to the day-to-day demands of the disease.

As we continue work towards treatments and a cure, our Peer-to-Peer Team is ready to talk! We are ready to listen and eager to share our own experiences with you. Although we are not healthcare professionals, we will try to direct you to the resources that you need to deal with issues around FSHD and to provide information about FSHD to your doctor.

Please call the FSH Society at (617) 658-7878 to be connected with a member of our Peer-to-Peer Team. A new friend will call you shortly. We look forward to hearing from you!

Peers: *If you are a patient or other family member who would like to join the Peer-to-Peer Team, please contact the FSH Society, (617) 658-7878.*

Combined Federal Campaign (CFC)

Are you a federal, postal or military employee?

Federal civilian, postal and military personnel can donate to the FSH Society through the Combined Federal Campaign during the campaign season, September 1 to December 15, 2009. **The FSH Society code is #10239.**

For more information about the CFC, you may go to the official website: <http://www.opm.gov/CFC/>

Freedom to travel with FSHD

By Howard L. Chabner

I am 52, have FSHD, began using an electric wheelchair in 1990, and have been unable to walk at all since 1996. Since 1996, my wife Michele DeSha, who is able-bodied, and I have traveled to France, Israel, Italy, Spain and Switzerland, besides many cities in the United States. We do our own planning and we travel on our own.

Travel in a wheelchair requires extra planning, patience and resourcefulness. (Of course it also helps to have an ideal traveling companion like Michele!) You must be prepared for frustrations and unexpected obstacles, but the highs are really, really high. Like everyone who enjoys traveling, the highs come from the beautiful and fascinating art, architecture and natural scenery one sees, the people one meets, the culture one experiences, the history one learns, and the food and drink one relishes. But as a disabled traveler, there are also some unique high points: the sense of empowerment and optimism one gets in being able to travel, in overcoming the obstacles and barriers, in seeing how other places handle access, and in noticing progress on repeat visits. Often the differences in access from one place to another yield interesting and valuable insights about cultural differences.

In some ways, using a wheelchair has enabled me to see and experience more than when I was a slow, precarious walker. Having returned to places I lived or visited when I walked—St. Louis, Tel Aviv, Boston and Rome, for example—I realize now that, even when I could walk a few miles a day, I was limited in what I could do. Now, although there are other obstacles, constant fear of falling, looking at the ground all the time, and fatigued legs are not among them. (Sometimes, after rolling/walking many miles together, Michele will ask “Howard is your thumb tired yet?”) And now I realize that places are much smaller than they seemed.



Howard in his wheelchair, on a lift in Capri, Italy

A great trip starts with meticulous research. Paradoxically, it's good research before your trip that enables spontaneity on the trip. I spend the most time researching transportation and hotel access. In recent years there have been many articles on the Internet about accessible travel to various destinations. We've contributed our share— please see our articles on Global Access News, a terrific accessible travel website—

www.globalaccessnews.com

I start our research there, and also simply Google “wheelchair access” and the name of the place we want to go. Next, I go on the official tourism websites of the places we are considering; many countries and cities have good access information on their websites. I've often emailed foreign tourism websites with specific questions and received informative responses in English. We also go on the websites of public transportation agencies to learn about bus, paratransit and train access.

As with all research, it's much better to use primary sources than to rely on secondary ones—for example, rather than relying on a travel agency's description of bus access, it's better to go to the transportation operator's website. Another good thing about researching access is that you also learn about the history, physical layout and culture of your destination.

Choosing the right hotel or apartment can make or break your trip. The two most important factors for us are location and quality of wheelchair access. A central location is absolutely critical; even a hotel with perfect access is undesirable if it isn't close to the sites you want to visit. Being in the center is more enjoyable and more efficient: you see more, you soak up more atmosphere and

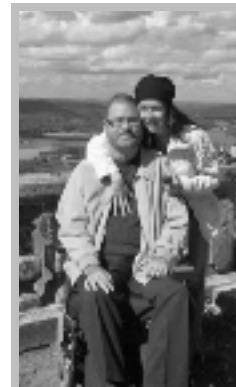
history, you can go back to your hotel and rest before dinner, and you can have a romantic dinner and stroll in the evening. Critically, being somewhere where you can roll/walk means you are much less dependent on transportation. Traveling when the weather is good also makes you less dependent on transportation.

In researching hotels abroad I start with regular guidebooks and Trip Advisor (www.tripadvisor.com), find well located hotels within our price range, and always send each hotel an access questionnaire I've developed and ask them to e-mail some photos of the bathroom. Many hotels are willing to e-mail photos. If you can walk, bathroom access is much less critical.

Transportation access is complex. By far the most difficult part of the trip is the plane ride, which could be the subject of a much longer article. Because of space limitations and because one has little or no control over air travel, I'll skip that and go on to ground transportation. Again, advance research is critical. When researching taxis, paratransit and even trains, it's important to tell the transportation provider the exact dimensions of your wheelchair—what is “wheelchair accessible”

depends a lot on the size of the wheelchair.

In cities in Europe and Israel we haven't rented a car, relying entirely on rolling/walking, public transportation and occasionally paratransit and taxis - a car is expensive, complex and unnecessary, and we don't want to add to pollution and congestion. We've had terrific experiences on public buses in Paris, which have lowered floors and accessible ramps, run frequently and on time, are spotlessly clean, and have skilled drivers and friendly, helpful fellow passengers. In Spain we were pleasantly surprised to find



Howard Chabner and Michele DeSha in the Perigord Region, France

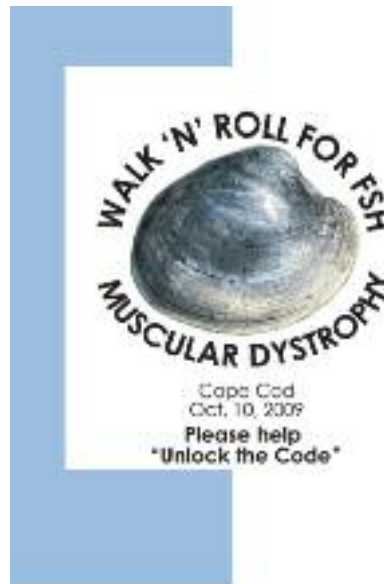
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Thank you Rich, friends on Cape Cod, and friends around the country

Rich Holmes, ably assisted by FSH Society Board member Robert Smith and wife, Patti, led a great new fundraiser for the Society on October 10. Rich will be happy to share his formula with others interested in putting a Walk 'n' Roll together.

Have you considered what you can do to help raise funds for the Society and FSHD research? In addition to the events already reported in *Watch* in recent months, these people have also put family and friends to work in other ways:

- ▶ Barbara and Robert Birnbaum asked friends to make contributions to the Society on the occasion of their 50th wedding anniversary
- ▶ Adi Segal and David Sullivan requested that friends make gifts in honor of their marriage
- ▶ Mary Rieger and Jude Watkins hosted a pig roast, with contributions coming to the Society



An Event to Benefit the
FSH Society



- ▶ Grace Corradino and friends held the annual Fire Island Yard Sale
- ▶ Wendy Herzberg hosted a Trunk Show in her New York apartment
- ▶ Toby and Leslie Berkeley included the Society as a recipient of funds raised at a Torah Dedication
- ▶ Alec Cohen carried out a recycling project for his Bar Mitzvah, sending the proceeds to the Society
- ▶ Marc Dunderman held a poker tournament to benefit the Society, in honor of an old friend's child.

Many of you are crafting letters to send to friends and family asking for gifts to support the FSH Society—the Kelly family, the Herzbergs, Howard Chabner, and Marshall Gillespie, to name a few. Max Teleki is making a presentation to a new prospect.

What will you plan to benefit the FSH Society in the coming months?

Freedom to travel with FSHD, *continued from page 11*

accessible and relatively plentiful taxis in the major cities—it was much easier to find an accessible taxi in Madrid than in New York and Boston.

In the U.S. we sometimes rent an accessible, lowered floor minivan from Wheelchair Getaways, a group of locally owned small businesses that operate in major cities.

www.wheelchairgetaways.com

Access at major museums, castles, antiquities, parks, churches and galleries is usually pretty good. These sites are often owned by the government or nonprofit organizations and are part of a country's patrimony and culture, so access is a priority.

And even if the access is only partial, a visit is still almost always worthwhile. Plus, it's important for Michele to see the



Howard finds a parking place in Italy.

sites even if I may not be able to. In Europe and Israel, all the government owned museums we've been to, and many of the others, offer free admission for a disabled person and a companion.

Museums and department stores often have good accessible restrooms, so if I am

near one, I often take the opportunity to visit the restroom.

It's also important to be prepared for wheelchair mishaps and medical

emergencies. For the former, research is key. I've required wheelchair repair (more than once) and a new battery charger in

Italy. I already had contact information for Quickie Italy, and they were able to help me find nearby wheelchair dealers, who fixed my chair quickly and professionally. I've needed minor medical care in Italy (including my first root canal); the U.S. Embassy and our hotels referred me to excellent English-speaking dentists and doctors. In Europe pharmacists can diagnose minor medical problems and dispense some medications without a doctor's prescription; Michele has had good experiences with pharmacists in Paris.

Traveling is one of the most enjoyable things in life, and one of the best learning and growth experiences. Although FSHD makes traveling more difficult, it need not prevent you from having superb travel adventures. Go!