

# FSH Watch

A publication of the FacioScapuloHumeral Society

## FSH Society Presents FY2004 Congressional Testimonies on FSHD Research

In 2003, we again went before Congress on behalf of all concerned with FSHD and submitted two testimonies before the United States House and United States Senate. The Society submitted Appropriations testimony before the Senate and House Appropriations Committee, Subcommittees on Labor, Health, Education and Related Agencies regarding appropriations for the National Institutes of Health (NIH) for research on Facioscapulohumeral muscular dystrophy on May 19, 2003 and May 23, 2003.

The Society again acquainted the members of the congressional committees with FSHD developments and the need for more funding on FSHD. The salient point of the testimonies is to express shock at the abysmal level of NIH funding on the FSHD disorder despite all of our effort. The testimony does not present extensive analysis and commentary on scientific opportunity or speculate on why funding is so inadequate. We asked the Congress to investigate why FSHD has been left behind in the broader picture of so many

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## What can YOU do?

By Judy Herzberg, Portland, Oregon

This past October, my husband and I attended the FSH Society's International Network Conference in Maryland. I approached the conference with fear. Seeing the effects of the disease on others causes me to project decline for my husband and son. For me, the hardest part has been worrying about the future. Seeing people in more advanced stages of FSHD gives me graphic details of what I fear most for our future. I avoid FSHD gatherings, yet need the information and support that only such a group can give. What surprised me about my experience at the conference was that on the other side of my fear, I found hope. I saw people living vital, rich lives despite the challenges brought by FSHD. I saw that people were making their ways through life, love, raising families, satisfying careers, and that while the bodies in the room may offer a glimpse into the decline for my husband and son in the future, I could also see that their lives could be full, which is all anyone can hope for.

I also found hope in research. Over the past decade, tremendous strides have been made toward understanding how FSHD works. Carol and Dan Perez have organized a very powerful group that works

to raise awareness about the disease and fund researchers working to understand FSHD. The support and cultivation of a network of research scientists studying the history and mechanisms of this disease is one of their most important achievements. Listening to the researchers present their findings, my husband and I became hopeful that as research and clinical trials continue, treatments will emerge. Funding is essential. I realized that the most important thing I can do for the health of my husband and son is to help raise funds for research.

The idea of asking people for money is uncomfortable for me. I first tested the waters by telling a few close friends about the conference and mentioning that Bill and I were considering writing a letter to friends and family asking them to support the FSH Society. I was surprised to get encouragement and offers of help.

My husband and I wrote letters about the FSH Society and the important work they are doing and sent it out to family and friends. Our friends helped us compose and print them. Since then, I have been overwhelmed by the outpouring of care that has come back to us. I feared

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*From the President***The FSH Society:****A Partnership of Patients, Families, Clinicians and Scientists**

As you read this newsletter, I hope that you are as pleased as I am with the extraordinary achievements of the Facioscapulohumeral (FSH) Society to date this year.

The Facioscapulohumeral Society (FSH Society) is a privately funded 501(c)(3) not-for-profit organization. From its inception, the FSH Society has been patient-driven. The Society was conceived in 1988 by two men affected by the disease, Dr. Stephen J. Jacobsen and me. The FSH Society helps families and friends bond with their fellow members both by their common knowledge of what it is to live with FSHD and by the ardent desire they all feel to be part of a concerted effort to discover how to treat the disease and, ultimately, to cure it.

As an FSHD sufferer myself for 41 years, I can tell you that this unbending adversity yields uncommon and unique strength. Advocate for yourself, others and the FSH Society. Read Judy Herzberg's article on how to make a difference. In our last newsletter, Ed Schechter called on all of us to get involved. It is up to you to advocate for yourselves now.

I accepted the confirmation by Secretary of Health and Human Services, Tommy G. Thompson, and Dr. Elias Zerhouni, Director, the National Institutes of Health (NIH) to serve as a member of the Muscular Dystrophy Coordinating Committee (MDCC), a federal advisory committee. The FSH Society worked very hard to bring about a carefully balanced law, the Muscular Dystrophy Community Assistance, Research and Education Amendments of 2001 (MD-CARE Act), to promote muscular dystrophy research. The goal then, as now, is to promote a significantly under-developed area of research – muscular dystrophy and in particular FSHD. The Society will continue to advocate for FSHD research.

We are making great progress despite the 40 years of historical neglect that

FSHD has endured. FSHD is yielding new and novel insights into DNA-to-DNA interactions and long-distance chromosome interactions among many other interesting areas of study. Scientists are very interested in the ramifications that this has on the paradigm of gene expression. Two more of our FSH Society fellows will shortly receive funding from the National Institutes of Health (NIH) for FSHD research.

The Society is working with physician-researchers, patients and pharmaceutical companies on clinical trials for FSHD. We are funding several projects to find compounds, agents and nutritional supplements to treat FSHD. The Society has initiated two projects to this end, under the directions of Dr. Sara Winokur, University of California and Dr. Graham Kemp, the University of Liverpool, United Kingdom. The NIH FSHD Registry at the University of Rochester is now online and connecting researchers who want to research FSHD with FSHD patients. Get involved in clinical trials and research by contacting the NIH FSHD Registry and tell them the FSH Society sent you.

We will follow this quarterly *FSH Watch Newsletter* with the annual *FSH Watch Research Report* that will have a comprehensive collection of research happenings and developments in FSHD from researchers and laboratories around the globe.

We need financial support to stay the course and grow. It is imperative that you contribute to our effort now more than ever. I count on you for your support and help at this critical time. Be very cognizant that unlike other non-profits who want regular support and donations, we actually and absolutely need funds to survive. The Society works diligently day and night to solve FSHD. It is your dollars at work for you!

Thank you for your past and continued support.

Daniel Paul Perez, President & CEO

## 2004 Congressional Testimonies, *continued from front page*

other areas of research at the NIH.

The entire written statement and testimony before Chairman Ralph Regula and the U.S. House of Representatives follows:

“Mr. Chairman, it is a great pleasure to submit this testimony to you today.

“My name is Daniel Paul Perez, of Lexington, Massachusetts, and I am testifying as President & Chief Executive Officer of the FacioScapuloHumeral Muscular Dystrophy Society (FSH Society, Inc.) and as an individual who has this devastating disorder.

“Facioscapulohumeral muscular dystrophy (FSHD) is the third most prevalent form of muscle disease. FSHD is a neuromuscular disorder that is transmitted genetically to 1/20,000 people. It affects up to 37,500 persons in the United States. FSHD can occur at any time by new mutations in the chromosome. Twenty-thirty percent of people affected by FSHD are believed to be new mutations.

For men and women, the major consequence of inheriting FSHD is progressive and severe loss of skeletal muscle. The usual pattern is of initial noticeable weakness of facial, scapular and upper arm muscles and subsequent weaknesses of other skeletal muscles. Retinal and cochlear disease, as well as mental retardation can be associated with FSHD although the pathogenesis and causative relationship to FSHD remains unknown.

FSHD wastes the skeletal muscles and gradually but surely brings weakness and reduced mobility. Many with FSHD are severely physically disabled and spend the last 30 years of their lives in a wheelchair. The toll and cost of FSHD physically,

emotionally and financially are enormous. FSHD is a life-long disease that has an enormous cost-of-disease burden and is a life sentence for the innocent patient and involved persons and their children and grandchildren as well.

“People who have FSHD must cope with continuing, unrelenting and never-ending losses. The most unlucky, those who are affected from birth, are deprived of virtually all the ordinary joys and pleasures of childhood and adolescence. No matter at which stage of life the disease makes itself known, there is never after that any reprieve from continuing loss of physical ability or ever, for a moment, relief from the physical and emotional pain that FSHD brings in its train. Every morning, FSHD sufferers wake up to face the reality that neither a cause for their disease nor any treatment for it has yet been found.

“FSHD denies a person the full range of choices in life. FSHD affects the way you walk, the way you dress, the way you work, the way you wash, the way you sleep, the way you relate, the way you parent, the way you love, the way and where you live, the way people perceive you, interact with you and treat you. You cannot smile, hold a baby in your arms, close your eyes to sleep, run, walk on the beach or climb stairs. Each new day brings renewed awareness of the things you may not be able to do the next day. This is what life is for tens of thousands of people affected by FSHD worldwide.

“Through the FSH Society, FSHD patients have found ways to be useful to medical and clinical researchers working on their disease. The FSH Society acts as a clearinghouse for information on the FSHD disorder and on potential drugs and

devices designed to alleviate its effects. It fosters communication among FSHD patients, their families and caregivers, charitable organizations, government agencies, industry, scientific researchers and academic institutions. It solicits grants and contributions from members of the FSH Society, and from foundations, the pharmaceutical industry, and others to support scientific research and development. It makes grants and awards to qualified research applicants. In less than six years, the FSH Society has raised more than \$1.1 million for research and has invested it in more than two dozen innovative research programs internationally. One of the FSH Society’s key assets, its Scientific Advisory Board, is composed of international experts whose awareness of current FSHD research ensures both that new research is not duplicative but complementary and that it will fill gaps in existing knowledge. The FSH Society’s work in education, advocacy, and training has led to increased funding in the United States and abroad. It was a key participant in drafting the Muscular Dystrophy Community Assistance Research and Education Act of 2001 (MD CARE Act) which in the United States mandates research and investigation into all forms of Muscular Dystrophy.

“A decade of progress in FSHD has led to the discovery of many novel genetic phenomena never seen before in human disease and genomics. Despite remarkable genetic insight and immense progress by a small team of scientists worldwide, the nature of the gene products remain enigmatic and the biochemical mechanism and cause of this common muscle disease remains unknown and elusive. The same is true for any treatment — none exist.

“More than a decade ago, we appeared before this Committee to testify for the first time. Ten years ago, I walked with some difficulty. Today, I sit in a wheelchair because of this disease called FSHD. Over the same ten years, the Appropriations Committees in both the U.S. House and the U.S. Senate have repeatedly instructed the National Institutes of Health (NIH) to enhance and broaden the portfolio in FSHD and muscular dystrophy in general.

*continued on page 4*

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 to its members and supporters. To be placed on the mailing list or to submit an article please write to: Carol Perez, *FSH Society*, 3 Westwood Road, Lexington, MA 02420. 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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**Elly Merkle, Carol Perez and Charles C. Perez**

## 2004 Congressional Testimonies, *continued from page 3*

The NIH accounting for the total overall NIH and the subset of muscular dystrophy appropriations in millions of dollars for the past five years is found in Table 1.

making it through the peer review process. FSHD grant reviews have been a constant source of frustration for FSHD researchers submitting grant applications to the

**Table 1 National Institutes of Health (NIH) Appropriations History**

Source: NIH/OD Budget Office & NIH CRISP Database On-line (Dollars in millions)

Fiscal Year	NIH Overall Dollars	MD Research Dollars	MD % of NIH	FSH Research Dollars	FSHD % of MD	FSHD% of NIH
1999	\$15,629M	\$16.7M	0.107%	\$0.4M	2.39%	0.0026%
2000	\$17,821M	\$12.6M	0.071%	\$0.4M	3.18%	0.0022%
2001	\$20,458M	\$21.0M	0.103%	\$0.5M	2.38%	0.0024%
2002	\$23,296M	\$27.6M	0.118%	\$1.3M	4.71%	0.0056%
2003	\$27,067M	\$31.4M	0.116%	\$1.5M	4.78%	0.0055%

“Despite major initiatives from the volunteer health agencies and the extramural community of researchers, FSHD research at the NIH and funding through the NIH is negligible in muscular dystrophy. Notwithstanding these positive changes at the NIH as well as major cooperative initiatives from the volunteer health agencies and the extramural community of researchers, we realize that major changes are slow but we are hopeful that this year the NIH will initiate new and increased funding for FSHD.

“Our concern is that the funding increases for facioscapulohumeral muscular dystrophy (FSHD) have been abysmal. In the last eighteen months, four grants directly and specifically pertaining to FSHD were submitted. One of the four, a small R21 style grant, was funded by the NIH Committee for Scientific Review (CSR). Despite the Congressional mandate to accelerate research on FSHD, FSHD grant applications are still not

program and review staff of CSR. Submitting R01, P01, R21 grant applications calls/contracts has been a frustrating and time consuming endeavor for most researchers in the FSHD community since it bears little fruit. (See Table 2.)

Since 1994, not a single R01 or P01 grant application focusing directly on a critical aspect of FSHD has survived the peer review process at CSR despite the high quality of researchers and the leading edge of scientific thought and opportunity involved with FSHD. We choose not to disagree with the peer review process at NIH nor do we seek to change the peer review requirement. However, we strongly emphasize that there is a shortage of reviewers with the required expertise to guarantee the review deserved by grant submissions in the muscular dystrophy area. Review of FSHD proposals, with novel genetic phenomena and mechanisms and a leading edge of scientific thought,

are particularly needful of that expertise. FSHD is unique among known (muscle) disorders in that the molecular mechanism is unprecedented in medical genetics. The chromosomal lesion in FSHD was detected more than 10 years ago. However, how the chromosomal rearrangement translates into FSHD symptoms and disease is almost completely enigmatic and involves novel molecular mechanisms. This should be addressed through multidisciplinary training, development and mentorship programs by the NIH. We also emphasize that the NIH must offer ways to ameliorate the difficulties in this aspect through targeted programs, training scientists and short- and long-term outreach efforts to produce the desired input to its own NIH process.

“Congress has been very generous with the NIH. Congress has repeatedly mandated more effort in muscular dystrophy research in general and FSHD research in particular. But this is not happening.

“We ask Congress to investigate why this is happening and request an explanation from the NIH accounting for the failure to do better in the area of FSHD despite repeated Congressional requests. Three R01 research grants funded on FSHD (two of them only peripherally FSHD-related at best) in four decades is not enough. We implore Congress to request the NIH to specifically build the research portfolio on FSHD through all available means, including re-issuing specific calls for research on FSHD at an accelerated rate, to make up for historical neglect.

“Mr. Chairman, we trust your judgment on the matter before us. We believe the Committee should explore why muscular dystrophy in general and FSHD in particular has been left behind in the great rise in research support at the NIH. Frankly, we are extremely frustrated that amid a huge increase in funding and strong unambiguous expressions of Congressional support, the NIH commitment in facioscapulohumeral muscular dystrophy (FSHD) is so weak. Only you can answer that question.

“Mr. Chairman, again, thank you for providing this opportunity to testify before your Subcommittee.”

**Table 2 National Institutes of Health (NIH) R21, R01, P01 Grants for FSHD**

Source: NIH CRISP Database On-line

Fiscal Year(s)	Total No. of FSHD P01 Grants	Total No. of FSHD R01 Grants	Total No. of FSHD R21 Grants
1972-1998	0	3	0
1999	0	1	0
2000	0	1	0
2001	0	1	0
2002	0	0	6
2003	0	0	7

***Your contribution to the FSH Society is tax-deductible and ensures the on-going work of YOUR advocacy group. Please send your donation now. Please see donation form on the back page.***

## What can YOU do? *cont. from front pg.*

that people would feel hounded by requests to support yet another worthy cause. Instead, friends and family members called to thank us for letting them know about the organization, offered to help, and suggested others who would be interested in receiving our letter. Our parents sent letters to their social circles. Friends mentioned being grateful that it was now out in the open; they could talk with us about it. We never knew that they felt they couldn't.

Our letter has been quite successful in terms of raising funds. The invaluable benefits that have come our way are to realize that we are not alone. We have generations of friends and family who care about us. We have let them in on how the disease has affected us, and they want to help us, and by connection, others with the same problem. I am glad to have reached out and showed our needs. I learned that there are so many ways to help. Friends found their own ways of joining our cause. It is not that I didn't know my friends and family cared; it is that they have broadened my notion of how to care. I've had offers to help stuff envelopes, to accompany me to doctors' appointments, to run fund-raisers, babysit, edit and print letters, to go for walks. I hope now I will be better about noticing when other people need help and finding my way to offer it.

In light of that, I offer my help to anyone wishing to contact and involve their friends and family. Those of us with FSHD in the family are a relatively small group. The most powerful hope we have is that we, along with those who know and care about us, will keep working to find a cure for FSHD and, one day, we will.

Sincerely, Judy Herzberg

*The letter from Judy and Bill Herzberg to family and friends appears at right.*

### How to Contact Us

Patients, professionals, & other parties interested in FSHD can contact us at

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12 December 2002

Dear Family and Friends,

We are writing to ask for your help.

Facioscapulohumeral muscular dystrophy (FSHD) is a disease that affects about 1 in 20,000 people worldwide and, as you know, has affected our family. It is a common, non-lethal form of muscular dystrophy whose major symptom is the progressive weakening and loss of skeletal muscles. The origin of the name: face (facio), shoulder girdle (scapulo) and upper arms (humeral) indicates the usual location of these muscular weaknesses. There is no treatment or cure. About a month ago we went to Maryland to attend a conference sponsored by the FSH Society so that we could learn more about the disease and find out what we could do to help others in the same situation. The FSH Society is a small group of 547 members, comprised almost exclusively of people with the disease, their families and friends. As often happens, the dedicated work of two people, Daniel and Carol Perez, was the catalyst for this organization which was created because of a need for a comprehensive resource for FSHD individuals and families. When a genetic marker for FSH was found in the early 1990s, the Society supported and encouraged scientific and clinical research through education, grants and private contributions. In 2001 the FSH Society lobbied for federal funds and helped pass the Muscular Dystrophy Community Assistance, Research, and Education Act (S.805). Recently, Daniel and Carol led the successful effort to get the National Institute of Health to list FSH and therefore make it eligible for federal research funds. These are great strides. The Society has also been instrumental in setting up a National Registry of patients with FSH in order to collect data about this registered group which helps speed up the time it takes to test potential treatments.

Here in Portland we are fortunate to have an FSH support group where we can go to share information and ask questions. One of the commitments we made after the conference in Maryland was to help organize an effort to reach doctors who may be treating FSH patients here in Portland and the Pacific NW region. Our goal is to expand these doctor's awareness of the existing support group, encouraging people with FSH to join and become documented, ultimately leading to more research which is vital for successful treatment of the disease.

We hope that you will join us in this effort by making a donation to the Research and Education Fund of the FSH Society. Eleven years ago a movement was started and we believe that eleven years from now your efforts will bear fruit. We thank you for whatever contribution you can make. —Judy & Bill

[Postscript by Bill Herzberg accompanying letter to family and friends]

As many of you know, I have FSHD, a hereditary progressive muscle disease which fortunately is less severe and less common than Duchenne's muscular dystrophy. What this means to me personally is that I can't raise my arms overhead, I have difficulty using my hands, I stumble a lot, and I anticipate further decline. Despite this, in many ways I feel strong. I recently completed a six day, 500 mile bike trip across Oregon with my sister Wendy, my cousins and friends.

FSHD gets little media attention and little funding for research from either the government or Jerry Lewis' Muscular Dystrophy Association. It's become clear that any meaningful efforts to overcome this condition will come from within - from patients, friends and families. Our numbers are small and, unfortunately, many of the people affected remain undiagnosed. Of the approximately 14,000 people with FSHD in the U.S., 570 are members of the FSH Society which Daniel Perez organized.

Daniel, a Harvard classmate, lived in my dorm and we were casual acquaintances. He wore orthotic leg braces and we both had funny gaits - but neither of us knew we shared the same diagnosis and similar fears about our futures. Dan was a neuro-biology major and I was a bio-chemistry major, both pre-med types. Daniel decided not to go the medical/graduate school route but rather to create an organization dedicated to finding a cure for FSHD. You have read in our letter about some of the achievements that Daniel and his mother Carol have accomplished. The support and cultivation of a network of research scientists is one of their most important achievements. These scientists, sponsored by the FSH Society, studied the natural history of the disease, have embarked upon clinical drug trials and have begun to unravel the disease's mechanism. Daniel has successfully lobbied Congress for NIH research dollars, not just for FSHD but for all muscle diseases. This will result in the establishment of three federally funded centers of excellence in muscle research. Eventually treatments will emerge and perhaps someday there will be a cure.

While Daniel has been mobilizing the FSH Society, I have been leading a full life and have had the luxury of avoiding the subject of FSH for years. Judy and I are happily married and I find plenty of time to enjoy our children, Bea and Harry. I work too hard, but I am able to work. I feel strong but worry about my future and I worry for my children. I envision a time in the not-too-distant future when I will be incapable of working as a physician and think about the possibility of retooling as a muscle scientist. And, most importantly, I am hopeful that real treatments and perhaps a cure will be available for future generations. Why so hopeful? Scientifically, the pace of understanding at the level of molecular biology is accelerating, and the funding for sustained research will soon fall into place.

Over the years friends and family have asked what they can do to help. Here's one way - donate to the FSH Society Research and Education Fund. Whatever you can send will make a difference. —Bill

## MD-CARE Act of 2001 Progresses Towards Research and Education Plan for Dystrophy

On May 7, 2003, Daniel Paul Perez, President and CEO, was confirmed to serve as a member of the Federal Advisory Committee called the Muscular Dystrophy Coordinating Committee (MDCC). The establishment of the MDCC was called for in the Muscular Dystrophy Community Assistance, Research and Education Amendments of 2001 (MD-CARE Act). Tommy G. Thompson, Secretary of Health and Human Services and Dr. Elias Zerhouni, Director, the National Institutes of Health (NIH) requested and reviewed nominations for the MDCC committee.

The Committee is asked to "coordinate research activities across the National Institutes of Health (NIH) and with other Federal health programs and activities relating to the various forms of muscular dystrophy, including Duchenne, myotonic, facioscapulohumeral muscular dystrophy (FSHD)," and six other forms of muscular dystrophy.

The law directs the Committee to develop a plan for conducting and supporting research and education on muscular dystrophy through the NIH and other federal national research institutes. The plan is to be submitted to Congress within the first year of the establishment of the MDCC.

### Abstract of Public Law 107-84:

"To amend the Public Health Service Act to provide for research with respect to various forms of muscular dystrophy, including Duchenne, Becker, limb girdle, congenital, facioscapulohumeral, myotonic, oculopharyngeal, distal, and Emery-Dreifuss muscular dystrophies.... The Director of NIH, in coordination with the Directors of the National Institute of Neurological Disorders and Stroke (NINDS), the National Institute of Arthritis and Musculoskeletal and Skin Diseases (NIAMS), the National Institute of Child Health and Human Development (NICHD), and the other national research institutes as appropriate, shall expand and intensify programs of such Institutes with respect to research and related activities concerning various forms of muscular dystrophy, including Duchenne, myotonic,

facioscapulohumeral muscular dystrophy (referred to in this section as 'FSHD') and other forms of muscular dystrophy."

The function of the MDCC will be to develop a plan for 'conducting and supporting research and education on muscular dystrophy' through the national research institutes, and shall periodically review and revise the plan. The plan shall

(a) provide for a broad range of research and education activities relating to biomedical, epidemiological, psychosocial, and rehabilitative issues, including studies of the impact of such diseases in rural and under served communities;

(b) identify priorities among the programs and activities of the National Institutes of Health regarding such diseases; and

(c) reflect input from a broad range of scientists, patients, and advocacy groups."

The structure of the MDCC is 15 members, including the Chair, appointed by Tommy Thompson, Secretary of Health and Human Services. Two-thirds of the members represent government agencies, including the directors or their designees of each of the national research institutes involved in research with respect to muscular dystrophy and representatives of all other Federal departments and agencies whose programs involve health functions and responsibilities relevant to such diseases. Included are the Centers for Disease Control and Prevention (CDC), the Health Resources and Services Administration (HRSA), and the Food and Drug Administration (FDA), Department of Education, Department of Defense (DOD) and others. The remaining one third of the members are a broad cross section of persons affected,

included or involved with muscular dystrophy e.g. patients, agencies, parents. The two-thirds, one-third composition of the committee was directly written into the MD CARE Act at the request of the FSH Society, Inc. since the first draft did not include the one-third public members. Again, the FSH Society ensured a comprehensive law that addresses the need for research for all of the major muscular dystrophies.

The FSH Society pioneered and laid the ground work for this law over a period of eleven years. In 2001, the Parent Project and the Muscular Dystrophy Association brought an enormous amount of energy to Washington, DC. This landmark legislation would not have been a reality without the unprecedented cooperation of all three organizations.

In the last six months, progress on the MD CARE Act of 2001 has been very apparent and there is momentum building for research and education for all nine muscular dystrophies including FSHD. We are delighted and honored that Daniel P. Perez was Secretary Tommy Thompson's choice to serve and contribute to this important task. The first meeting is in Bethesda, Maryland in July 2003. We will provide updates, hyperlinks, and information from the MDCC to you when released and as permitted.

Note: A complete copy of the MD-CARE Act can be found online at the Thomas Locator:

(<http://thomas.loc.gov/home/thomas.html>)  
Once there, click on "Public Laws by Law Number" under the Legislation link. That will take you to the 107th Congress' public laws. Click on Public Laws 107-51 - 107-100. Then scroll down to Law 84 (H.R. 717). You will find a link to bring up the text or a printable pdf file.

***Do you have a few hours to give to the Society's work? Many things can be done right from where you live. We need you!!***

## Timeline: Progress in FSHD — 2003

- **January 4, 2001**

NIH issues PAS-01-041 Therapeutic and Pathogenic Approaches for the Muscular dystrophies PAS-01-041. The funding mechanism is research project grant (R01) and the program project grant (P01). The NIH has earmarked approximately US\$5.0 million in total costs during fiscal years 2002- 2004. See NIH web site:

[http://www.niams.nih.gov/rtac/funding/grants/pa/pas\\_01\\_041.pdf](http://www.niams.nih.gov/rtac/funding/grants/pa/pas_01_041.pdf)  
(Please see article on page 10 in this newsletter.)

- **January 14, 2003**

The NIH (NIAMS, NICHD, NINDS) holds and conducts an informational pre-application meeting at which Program and review staff make presentations and explain their goals and objectives for the Muscular Dystrophy Cooperative Research Centers described in RFA-AR-03-001.

- **January 21-22, 2003**

The NIH (NIAMS, NINDS, and NICHD) convenes the Muscular Dystrophy Research Task Force to identify ways to increase the level of understanding of muscular dystrophies and promote clinical treatment and therapy. A summary of the second meeting of the Task Force is not currently available from the NIH or on its web site. Daniel Paul Perez participated as a member of the Task Force on behalf of the FSHD community. (Please see article on page 18 in this newsletter.)

- **February 13, 2003**

The FSH Society, Inc. re-opens dialogue with the Muscular Dystrophy Association Canada (MDAC) and the Canadian Institutes of Health Research (CIHR) on FSHD research programs in Canada. Contacts and discussion are ongoing with Director, Programs and Services, and Executive Director of the MDAC. FSHD is the fourth largest patient constituent group in the MDAC database registry (485 persons) of nearly one hundred disorders and MDAC confirms that there is not one single research project on FSHD in Canada.

- **February 24, 2003**

The NIH receives applications for contract RFA AR-03-002 for developmental

planning for Muscular Dystrophy Cooperative Research Centers. RFA uses R21 Grant award mechanism and will award approximately US\$1 million for five grants for two years. See NIH site:

<http://grants.nih.gov/grants/guide/rfa-files/RFA-AR-03-002.html>

- **February 24, 2003**

The NIH receives applications for contract RFA AR-03-001 on Muscular Dystrophy Cooperative Research Centers. RFA uses U54 Grant award mechanism and will award approximately US\$4.5 million for five years to be distributed to three or four CORES. See NIH site:

<http://grants.nih.gov/grants/guide/rfa-files/RFA-AR-03-001.html>

- **February 27, 2003**

The NIH Office of Rare Diseases (ORD) and the National Center for Research Resources (NCRR) issues a Request for Applications (RFA: RR-03-008) for a Rare Diseases Clinical Research Network. RFA uses U54 grant award mechanism RFA is a one-time solicitation. The anticipated award date is September 28, 2003.

Approximately \$7.0 M in FY 2003 to fund 4 RDCRCs (\$5 M) and 1 DTCC (\$2 M). An applicant must request a project period of five years. See NIH site:

<http://grants1.nih.gov/grants/guide/rfa-files/RFA-RR-03-008.html>

- **April 29, 2003**

The NIH Office of Rare Diseases (ORD) and the National Center for Research Resources (NCRR) receives applications for RFA: RR-03-008 for a Rare Diseases Clinical Research Network. The

anticipated award date is September 28, 2003. See February 27, 2003 above for more information.

- **May 7, 2003**

Secretary of Health and Human Services Tommy G. Thompson and Dr. Elias Zerhouni, Director, the National Institutes of Health (NIH) confirm Daniel Paul Perez to serve as a member of the Federal Advisory Committee called the Muscular Dystrophy Coordinating Committee (MDCC). The establishment of the MDCC was called for in the Muscular Dystrophy Community Assistance, Research and Education Amendments of 2001 (MD-CARE Act 2001.) (Please see article on page 6 in this newsletter.)

- **May 19, 2003**

The FSH Society presents U.S. Senate testimony submitted in written format before Senators Specter and Harkin. The complete testimony is identical to the U.S. House testimony given on May 23, 2003. Please check FSH Society Internet web site, or contact the FSH Society for a written copy.

- **May 23, 2003**

The FSH Society presented written testimony before the U.S. House Committee on Appropriations, Subcommittee on Labor, HHS, Education and Related Agencies, chaired by Representative Ralph Regula. To view the complete testimony, please check the FSH Society web site, or contact the FSH Society for a written copy. (Please see article on front page of this newsletter.)

## Statement by Stephen I. Katz, M.D., Ph.D., Director, NIH/NIAMS before Congress

On April 8, 2003, Stephen I. Katz, M.D., Ph.D., Director, National Institute of Arthritis and Musculoskeletal and Skin Diseases (NIH/NIAMS) appeared as a witness before the Senate Subcommittee on Labor-HHS-Education Appropriations to provide the Fiscal Year 2004 Budget Request. We were delighted that Dr. Katz mentioned the valuable work on FSHD supported by the NIAMS as well as the work of the various task force, committees and public/private partnerships. Dr. Katz's reference alludes to the study by Drs. Rick Lemmers, de Kievit, Sandkuijl, George W.A.M. Padberg, GJ van Ommen, Rune Frants, and Silvere van der Maarel of the Department of Human Genetics, Center for Human and Clinical Genetics, Leiden University Medical Center, Leiden, The Netherlands titled: "*Facioscapulohumeral muscular*"

*continued on page 8*



## Statement by Stephen I. Katz, *continued from page 7*

*dystrophy is uniquely associated with one of the two variants of the 4q subtelomere.*" Nature Genetics 2002 Oct; 32(2):235-6. This Pubmed Medline abstract for this paper states: "Contractions in the polymorphic D4Z4 repeat array of subtelomere 4qter cause autosomal dominant facioscapulohumeral muscular dystrophy in humans. A polymorphic segment of 10 kb directly distal to D4Z4 exists in two allelic forms, 4qA and 4qB. Although both alleles are equally common in the general population, we now report that FSHD is associated solely with the 4qA allele." The NIAMS funded this research and continues to fund it under a R21 style grant numbered AR48327-01.

For the entire testimony please see the hyperlink at the NIH/NIAMS website:

[http://www.niams.nih.gov/ne/reports/congree\\_rep/cj2004/open2004.htm](http://www.niams.nih.gov/ne/reports/congree_rep/cj2004/open2004.htm)

Selected excerpts from Dr. Katz's testimony follow:

"Mr. Chairman and Members of the Committee:

"I am pleased to present the President's budget request for the National Institute of Arthritis and Musculoskeletal and Skin Diseases (NIAMS). The fiscal year (FY) 2004 budget includes \$502.778 million, an increase of \$17.005 million over the FY 2003 enacted level of \$485.773 million comparable for transfers proposed in the President's request.

"The budget increases over the last few years have made a tremendous difference in the studies we have been able to launch, particularly in clinical research including clinical trials in a wide variety of diseases as well as the expansion of vital scientific infrastructure in a creative way ...

"One of the priority areas in the new NIH Roadmap Initiative is the development of public/private partnerships. The NIAMS has had a number of positive experiences in this area, and I will mention two ongoing examples ...

"The second ... The NIH has been actively engaged in implementing the mandates of the MD-CARE Act, and has worked closely with representatives of the muscular dystrophy (MD) research and patient communities in this effort.

Specifically, the NIAMS, NINDS, and NICHD have partnered to issue new research solicitations for MD cooperative research centers, and for developmental planning grants for future centers. In addition, we are developing an initiative to support the training of basic and clinical researchers to study muscular dystrophy. To underscore the importance of stimulating and supporting further work in this area, the NIH has established an MD Research Task Force, which includes NIH scientific staff, as well as researchers, clinicians, and patient representatives. This group will help ensure that we pursue all promising opportunities to boost MD research and training, and it will also complement the work of the newly established inter-agency Muscular Dystrophy Coordinating Committee, which was called for in the MD-CARE Act . . .

"One of the most active and productive areas within the Institute's research portfolio is in the muscular dystrophies – a group of genetic diseases characterized by progressive weakness and

degeneration of the skeletal or voluntary muscles which control movement. Research advances from NIAMS investments in this area include: (1) the finding that people with facioscapulohumeral muscular dystrophy (FSHD) have an exclusive association with one of the two different forms of the chromosomal region linked to the disease. This work may lead to a better understanding of the instability of the genetic locus associated with FSHD. . .

"We are proud of the advances that scientists supported by the NIAMS have achieved and we are excited about initiatives that we have launched. Patients and their families are looking to us with hope and anticipation for answers to what causes their diseases, as well as how their diseases can be better treated and even prevented. We are confident that public health in general as well as daily life for affected individuals in particular will benefit from NIAMS research in the extensive and diverse array of chronic diseases within our mission areas of bones, joints, muscles, and skin."

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## About the FSH Society

The Facioscapulohumeral Society (FSH Society) is an independent, 501(c)(3) non-profit and tax-exempt U.S. corporation organized to address issues and needs specifically related to Facioscapulohumeral Muscular Dystrophy (FSHD). Papers certifying its incorporation, bylaws and tax-exempt status are deposited at the corporation's east and west coast offices and the office of its General Counsel in Washington, D.C.

FSHD is a muscle disease with a frequency in the population of between 4 and 10 per 100,000. The disease is inheritable and the responsible gene may be located on chromosome 4. The expression of symptoms requires inheritance of the defective gene from only one affected parent, and an individual of either sex has a fifty percent chance of inheriting the gene from that affected parent. The major consequence of inheriting the disease is that of a progressive loss of skeletal muscle, with a usual pattern of initial noticeable weakness of facial, scapular and upper arm muscles and subsequent developing weaknesses of other muscles of the torso and lower limbs. Early facial weaknesses often provide a clue to the physician that distinguishes this disease from other neuromuscular diseases that can be similar in appearance.

The age of onset is variable as is the eventual extent and degree of muscle loss, but noticeable muscle weaknesses are usually present by the age of twenty and are recognizable in all but a small percentage of adults who carry the gene. The prognosis includes both a loss of muscular strength that limits personal and occupational activities of most FSHD individuals and a loss of mobility in perhaps twenty percent of the cases. Hearing loss and retinal abnormalities associated with FSHD have been reported, but the frequency of these effects and their relationship, if any, to the causative gene for the muscle defect are

*continued on page 9*



## Statement by Audrey S. Penn, M.D., Acting Director, NIH/NINDS before Congress

On April 8, 2003, Audrey S. Penn, M.D., Acting Director, National Institute of Neurological Disorders and Stroke (NIH/NINDS) appeared as a witness before the Senate Subcommittee on Labor-HHS-Education Appropriations to provide the Fiscal Year 2004 Budget Request. We are delighted that Dr. Penn mentions the valuable work on FSHD supported by the NINDS. Dr. Penn's reference alludes to the study by Drs. Rossella Tupler, Davide Gabellini and Michael Green of the University of Massachusetts Medical School (UMMS) Programs in Gene Function & Expression and Molecular Medicine titled "Inappropriate gene activation in FSHD: A repressor complex binds a chromosomal repeat deleted in dystrophic muscle," Cell 2002 Aug 9;110(3):339-48. This paper shows that a genetic defect called "deletion" of certain repetitive DNA sequences in people with FSHD allows nearby genes to go into overdrive. The NINDS funded this research and continues to fund it as well under a R21 style grant numbered NS43973-01.

For the entire testimony please see the hyperlink at the NIH NINDS website:

[http://www.ninds.nih.gov/about\\_ninds/FY\\_2004\\_congressional\\_testimony.htm](http://www.ninds.nih.gov/about_ninds/FY_2004_congressional_testimony.htm)

Selected excerpts from Dr. Penn's testimony follow:

"I am Audrey Penn, Acting Director of the National Institute of Neurological Disorders and Stroke (NINDS). I am

pleased to present the President's budget request for NINDS for Fiscal Year 2004. The fiscal year (FY) 2004 budget includes \$1,469 million, an increase of \$13 million over the FY 2003 enacted level of \$1,456 million comparable for transfers proposed in the President's request ...

"The mission of NINDS is to reduce the burden of neurological disorders; that is, the many diseases that affect the brain, spinal cord, muscles, and nerves of the body. Today I will touch on these points and concentrate on what NINDS is doing to expedite progress ...

"A few findings from the past year illustrate this progress: Scientists studying genes ... found clues about how a chromosome defect causes facioscapulohumeral dystrophy, a common form of muscular dystrophy...

"Neurological disorders have always challenged the best efforts of medicine. The intricacy of the brain is awesome, its workings are elusive, and an extraordinary variety of disorders affect the nervous system. Furthermore, the brain and spinal cord are difficult to access, sensitive to intervention, and reluctant to regenerate following damage. However, building on advances in basic science, progress is improving peoples' lives, and prospects for the future are even more encouraging. We are working to engage the best minds in the nation and provide them with the resources they need to devise ways to prevent, treat, or, ultimately, cure neurological disorders. Thank you."



***Your contribution to the FSH Society is tax-deductible  
and ensures the on-going work of YOUR advocacy  
group. We need your continued support.***

***Please send your donation now.***

***The donation form can be found on the back page.***



## About the FSH Society,

*continued from page 8*

uncertain.

The FSH Society was created because of a need for a comprehensive resource for FSHD individuals and families. Purposes of the organization are:

- to encourage and promote scientific and clinical research and development through education of the general public, governmental bodies and the medical profession;
- to support such research and development through solicitation of grants and contributions from private foundations, the pharmaceutical industry and others;
- to accumulate and disseminate information about FSHD;
- to actively cooperate with related organizations and foster communication among all interested parties;
- to represent individuals and families with FSHD.

The Society invites contact from any interested individuals, families, physicians, caregivers, charitable organizations, government agencies, industry, scientific researchers and academic institutions. Any inquiries regarding membership, charitable donations, purposes and goals or other issues pertaining to the Society and FSHD, should be addressed to the east or west coast offices.

### **The Combined Federal Campaign #2662 for the FSH Society, Inc.**

Federal employees and military personnel can donate to the FSH Society, Inc through the Combined Federal Campaign (CFC). Please consider making a contribution to the FSH Society through the CFC.

The CFC is operated by the United States Government Office of Personnel Management (OPM). The FSH Society, Inc. CFC code is #2662. For more information about the CFC you may visit the OPM website at

<http://www.opm.gov/cfc/index.htm>.

## NIH Continues Program Announcement for Therapeutic and Pathogenic Approaches for the Muscular Dystrophies

The NIH has an ongoing program announcement with funds set aside, a PAS, called Therapeutic and Pathogenic Approaches for the Muscular dystrophies PAS-01-041. This PAS was a renewal of the PA offered in 1998 PA-98-044 and hopefully, after three years, the new PAS will be renewed. The FSH Society was instrumental in bringing about the prior 1998 announcement after many years of work in Washington, DC and at the NIH. The mechanisms of support will be the individual research project grant (R01) and the program project grant (P01). The NIAMS and NINDS intend to commit approximately US\$5.0 million in total costs to fund competing applications submitted in response to this announcement during fiscal years 2002-2004. **Please note:** October 1, 2003 will be the last opportunity to submit a grant application under this program.

Several examples of research requested for study under this announcement are:

- Determine basis of differential involvement of muscles, reflected by the regional pattern of disease. Comparison of muscle groups might show the cause of relative specificity of affected muscles. Comparing expression patterns of RNA and protein in affected and non-affected muscle will provide insights into

alterations occurring as the disease progresses.

- Explore the role of inflammation in FSHD. While FSHD has been described as the most inflammatory form of muscular dystrophy, there is no evidence that disease severity is lessened by administration of the anti-inflammatory drug prednisone. It is necessary to explore the relationship between inflammatory cells, muscle cell death, and blood vessels.

- Study properties of muscle cells derived from affected tissue. Cells cultured from FSHD muscle show increased sensitivity to oxidative stress. This needs to be followed up by studies verifying that this occurs in vivo and establishing how this cellular phenotype develops.

### THERAPEUTIC AND PATHOGENIC APPROACHES FOR THE MUSCULAR DYSTROPHIES

Release Date: January 4, 2001

PA NUMBER: PAS-01-041

National Institute of Arthritis and Musculoskeletal and Skin Diseases

National Institute of Neurological Disorders and Stroke

PAS uses R01 and P01 Grant award mechanisms

[http://www.niams.nih.gov/rtac/funding/grants/pa/pas\\_01\\_041.pdf](http://www.niams.nih.gov/rtac/funding/grants/pa/pas_01_041.pdf)

## FSH Society Seeks Director of Development, Membership and Public Relations

We are searching for a professional with 5-10 years experience in non-profits in public relations, membership development, all aspects of fundraising and planned giving. Part-time salaried position two-three days per week. Please send inquiries and resume or C.V. to the FSH Society, Inc.

### The New York Community Trust Foundation Grant to the FSH Society, Inc.

Since 2000, at the request of an Anonymous Donor, The New York Community Trust awarded an annual grant of \$50,000 for five years to the FSH Society, Inc. The Society is grateful for the continued generosity and respect demonstrated by this gift. We express our gratitude to our Anonymous Donor.

Thanks to our Anonymous Donor and the New York Community Trust, these funds provide the resources to develop our organization into a responsive Society.

## Visit us @ [www.fshsociety.org](http://www.fshsociety.org)

By Daniel Paul Perez, President & CEO, FSH Society

Visit us and bookmark the FSH Society at its online Internet location [www.fshsociety.org](http://www.fshsociety.org)

The web site is still going strong and we are seeing a tremendous increase in both domestic and international traffic. We are getting the word out about FSHD! The home page at [www.fshsociety.org](http://www.fshsociety.org) contains a rich resource of material for those interested in FSHD.

For those not familiar with the site, the FacioScapuloHumeral Muscular Dystrophy Society home page contains information on the following: the FSH Society; FacioScapuloHumeral Muscular Dystrophy (FSHD); the FSH Society Online bulletin board and chat room; and previous FSH Society publications and information.

The chat room, hosted by Paul Closson, meets every Sunday 2 p.m. and 9 p.m. Eastern Time zone.

Professional Web designers and Web architects who would like to volunteer their time and services are encouraged to contact us. We fully appreciate the contributions and donations made to date to the Society to support this important and timely resource. Please consider making a donation to the FSH Society Internet fund.

We look forward to seeing you on-line!

## Wanted: Web Developers

The FSH Society continues to seek professional web developers or design firms interested in volunteering time and resources to update and redesign the Society's web site. Please address inquiries to the FSH Society, Inc.

## FSH Society Needs Volunteers!

The FSH Society is looking for marketing and fund-raising people to continue its work. Please contact Carol Perez, FSH Society Executive Director, at (781) 860-0501 if you have experience in these areas and would like to help with our work to solve FSHD!

## Clinical Activity

### Update: Albuterol/Oxandrolone Clinical Trial

June 2003: Unfortunately, the start of the clinical trial of albuterol and oxandrolone in FSH muscular dystrophy being performed by the Myopathy Study Group is still delayed. However, the problems relating to lack of availability of the long acting albuterol preparation to be used in the study as well as several other issues have been and are quickly being resolved.

Currently, John T. Kissel, M.D., Professor of Neurology, Ohio State University is hoping to have the study up and running after several last approvals and sign offs from companies and agencies involved. All of the members of the Myopathy Study Group remain committed to the successful completion of this study. For more information on albuterol trials please see previous issues of the *FSH Watch*.

### NIH National Registry of Myotonic Dystrophy and Facioscapulohumeral Muscular Dystrophy Patients and Family Members is Enrolling Individuals

By Daniel Paul Perez, FSH Society, Inc.

The University of Rochester Medical Center has been funded by the National Institutes of Health to establish a Registry of patients who have been diagnosed with myotonic dystrophy (DM) and facioscapulohumeral muscular dystrophy (FSHD). The Registry establishes and maintains a comprehensive list of individuals diagnosed with either of the two neuromuscular disorders. Participants will fill out a questionnaire to be entered into a data bank. No blood or tissue samples are needed. The Registry will assist researchers, from all over the United States, find and recruit individuals willing to participate in research studies. Currently, the registry is enrolling participants from the United States. For information about FSHD patient participation in the Registry contact: University of Rochester, National Registry of DM and FSHD Patients, 601 Elmwood Avenue, Box 673, Rochester, NY 14642-8673. Toll Free: 1 (888) 925-4302, Local (585) 506-0004, Fax: (585) 273-1255.

**e-mail:**

dystrophy\_registry@urmc.rochester.edu

**web:**

<http://www.dystrophyregistry.org>

**or**

<http://www.urmc.rochester.edu/nihregistry/contact.htm>

**Note:** Tell them we sent you!

### Brain and Tissue Banks Enlisting Registered Donors

The Brain and Tissue Banks for Developmental Disorders at the University of Maryland in Baltimore and the University of Miami are tissue resources established to further research aimed at improved understanding, care and treatment of developmental disorders. The Brain and Tissue Banks serve as intermediaries between people who wish to have tissue donated for research upon the time of their death and the researchers who need this tissue for their vital work. If you are interested in becoming a registered donor, or if you have any questions or concerns regarding the donation process, please contact Christine Wade, Project Coordinator. Thank you for taking the time to consider the possibilities offered to humanity through the gift of tissue donation. Internet site: [www.btbank.org](http://www.btbank.org)

Christine Wade, Project Coordinator, Brain and Tissue Bank, University of Maryland, 655 W. Baltimore Street, BRB 10-027, Baltimore, Maryland USA  
Phone: (800) 847-1539 or (410) 706-1755

### Participants needed for FSHD Muscle Biopsy Study

By Lynn Cos, RN, CCRC

Researchers at the University of Rochester Medical School are looking for individuals with FSHD interested in participating in a research study to further evaluate the gene causing FSHD. The study involves a one time visit to obtain a blood sample and a muscle biopsy. Researchers: Rabi Tawil, M.D. Contact: FSHD Tissue Bank Study, 601 Elmwood Avenue, Box 673, Rochester, NY 14642. Telephone: 1 (585) 275-6372.

### Current Happenings: Clinical Research at University of Liverpool, UK

Dr. Kemp (UK), one of two new Research Fellows, has contributed the following article to help our readers understand more about his work on FSHD. Please see page 18 for an article from our other new Research Fellows, Dr. Marahrens/Dr. Embade.

#### From the University of Liverpool, United Kingdom

By Graham Kemp, M.D., University of Liverpool, United Kingdom

I and my three colleagues in Liverpool, UK, are pleased to have been awarded the first FSH Society Roberts Foundation Nutrition Research Grant (FSHS-SMRF-01), for a project entitled "*Muscle damage by reactive oxygen species, muscle atrophy and effects of creatine supplementation in FSH muscular dystrophy.*"

A port city on the northwest coast of England, Liverpool was the European end of the transatlantic sea route to the USA, and may be known to some readers for its contributions to pop music in the 1960s. It is home to a large civic university with a thriving medical school, and to some of the country's biggest and busiest hospitals.

Three of the research team are based in the Faculty of Medicine of the University of Liverpool. I am in the Department of Musculoskeletal Science. Having trained in medicine at Oxford and specialized in clinical biochemistry, I held research positions at the Universities of Sheffield and Oxford before moving to Liverpool in 1996, where I am also Faculty Director of Postgraduate Research. My main research interests are in muscle biochemistry and muscle diseases and their study by non-invasive magnetic resonance techniques, and I take part in the outpatient clinical care of patients with muscular dystrophies.

Malcolm Jackson is Professor of Cellular Pathophysiology in the Department of Medicine. After training in biochemistry in Surrey and London, he held research positions in London before

*continued on page 14*

## Budget Numbers for Facioscapulohumeral Muscular Dystrophy

### National Institutes of Health (NIH) Appropriations History Budget Numbers for Facioscapulohumeral Muscular Dystrophy

Source: NIH/OD Budget Office & NIH CRISP Database On-line  
(Dollars in millions)

	FY 1987	FY 1988	FY 1989	FY 1990	FY 1991	FY 1992	FY 1993	FY 1994	FY 1995	FY 1996	FY 1997	FY 1998
NIH	Actual	Actual	Actual	Actual	Actual	Actual	Actual	Actual	Actual	Actual	Actual	Actual
Total (\$M)	<b>0.2</b>	<b>0.2</b>	<b>0.2</b>	<b>0.2</b>	<b>0.3</b>	<b>0.3</b>	<b>0.4</b>	<b>0.5</b>	<b>0.5</b>	<b>0.4</b>	<b>0.5</b>	<b>0.5</b>

## Budget Numbers for Total Muscular Dystrophy

### National Institutes of Health Appropriations

Source: NIH/OD  
(Dollars in millions)

Participating ICs	FY 1987 Actual	FY 1988 Actual	FY 1989 Actual	FY 1990 Actual	FY 1991 Actual	FY 1992 Actual	FY 1993 Actual	FY 1994 Actual	FY 1995 Actual
NHLBI	\$0.0	\$0.0	\$0.0	\$0.0	\$0.0	\$0.0	\$0.0	\$0.0	\$0.0
NINDS	2.4	2.6	3.7	4.7	5.6	5.6	7.1	7.1	
NICHD	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	
NEI	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	
NIA	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	
NIAMS	1.3	1.4	1.0	0.8	1.2	1.1	0.9	1.1	
NIMH	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	
NHGRI	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	
NCRR	0.9	0.4	0.9	0.0	0.0	0.4	0.4	0.6	
FIC	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	
NIH *	4.6	4.3	5.6	5.6	6.8	7.2	8.4	8.9	

\*May not add due to rounding.

by

### ons ar Dystrophy

FY	FY	FY	FY	FY	FY
1999	2000	2001	2002	2003	2004
Actual	Actual	Actual	Actual	Est.	Est.
0.4	0.4	0.5	1.3	1.5	

### Budget Numbers for Total NIH

#### National Institutes of Health Appropriations History

Source: NIH/OD Budget Office  
(Dollars in thousands)

Fiscal Year	Appropriation*
1987	\$6,182,910
1988	6,666,693
1989	7,144,764
1990	7,576,352
1991	8,276,739
1992	8,921,687
1993	10,326,604 1/
1994	10,973,653
1995	11,299,522
1996	11,927,562
1997	12,740,843
1998	13,674,843
1999	15,629,156 2/
2000	17,820,584
2001	20,458,130 3/
2002	23,296,382
2003	27,066,782

\*Amounts reflect enacted supplementals, rescissions and reappropriations.

1/ Includes appropriations totaling \$1,164 million for ADAMHA institutes following the ADAMHA Reorganization Act (42 USC 201) July, 1992.

2/ Beginning in FY 1999, includes funds available for diabetes research in accordance with the Balanced Budget Act of 1997.

3/ Beginning in FY 2001, VA/HUD began appropriating Superfund Research funds directly to NIEHS.

### ar Dystrophy Across NIH Institute

#### History for All Types of Muscular Dystrophy

OD Budget Office  
(in Millions)

1995	FY 1996	FY 1997	FY 1998	FY 1999	FY 2000	FY 2001	FY 2002	FY 2003	FY 2004
Actual	Actual	Actual	Actual	Actual	Actual	Actual	Actual	Estimate	Estimate
\$0.0	\$0.0	\$0.0	\$0.0	\$0.0	\$0.0	\$1.0	\$1.1	\$1.2	\$1.2
7.5	8.1	9.4	9.5	8.7	4.9	8.2	9.8	12.3	12.6
0.0	0.0	1.0	0.9	1.0	1.2	0.5	0.6	0.6	0.6
0.0	0.0	0.0	0.4	0.3	0.2	0.0	0.0	0.0	0.0
0.0	0.0	0.0	0.9	0.9	0.9	1.3	1.3	1.3	1.3
2.3	2.4	3.0	3.7	5.4	4.8	9.2	11.1	12.0	12.5
0.0	0.0	0.0	0.0	0.0	0.2	0.0	0.0	0.0	0.0
0.0	0.0	0.0	0.0	0.0	0.0	0.3	2.3	2.4	2.5
0.7	0.3	0.3	0.3	0.3	0.4	0.4	1.4	1.6	1.7
0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0	0.0
10.5	10.8	13.7	15.7	16.7	12.6	21.0	27.6	31.4	32.3

## Clinical Activity

### Current Happenings: Clinical Research at University of Liverpool, UK, *continued from page 11*

moving to Liverpool in 1984, where he is a former Dean of the Faculty of Medicine. Professor Jackson has a longstanding research interest in mechanisms of cell damage in muscle disease and ageing, particularly in the role of “reactive oxygen species” (of which more below).

Neil Roberts is Director of the University’s Magnetic Resonance and Image Analysis Research Centre (MARIARC), which applies magnetic resonance methods to research mainly in neuroscience. After training in physics in Liverpool, Cardiff and Aberdeen, Professor Roberts held research positions at the University of Durham and the University of California at Santa Barbara before moving to Liverpool in 1991, where one of his research interests is the mathematics of measuring body composition and structure. He is currently overseeing the expansion of the facilities at MARIARC with the purchase of two new magnetic resonance scanners.

The fourth member of the team is Bryan Lecky, Consultant Neurologist at our specialist neuroscience hospital, the Walton Centre for Neurology & Neurosurgery, Liverpool. After medical training at Cambridge and London, Dr Lecky held clinical and research positions in London before moving to Liverpool in 1987. His particular clinical expertise is in neuromuscular diseases, and he has extensive experience in clinical trials.

This eighteen-month project, which started on 1st January this year, combines three of our main research interests: quantitative magnetic resonance methods, mechanisms of muscle damage, and the therapy of muscle disease. It is a pilot study designed to investigate two questions: how tissue damage occurs in the muscle of patients with FSHD, and whether adding a naturally-occurring substance called creatine to the diet can improve muscle strength and reduce tissue damage.

The project arose from two lines of evidence. The first was work on creatine supplementation. Creatine is found mainly

in the muscle, where it plays a role in energy storage and use, and this makes it a popular dietary supplement among athletes. There has been a lot of interest in possible use in therapy and it is currently under trial in a number of different diseases.

Studies in mixed groups of dystrophies (including some patients with FSHD) have found short-term improvements in strength and daily-life activities. However, this is not well established for FSHD itself and there is a lot of argument about how it might work.

The second line of evidence is from recent work, funded by the FSH Society, showing that muscle cells in FSHD are particularly susceptible to a kind of damage called “oxidative stress,” which is caused by chemicals, produced in the body, called reactive oxygen species (Barrett, Tawil, Griggs & Figlewicz, ‘FSHD myoblasts possess reduced resistance to oxidative stress’ in *FSH Watch*, Spring 2001, pg. 70). The two lines of evidence are connected, as there is evidence that creatine supplementation protects against damage by reactive oxygen species, although how this might work is also controversial.

We are recruiting patients with FSHD from our clinic. To make sure that any effects we discover are specific, we will also study a smaller number of patients with myotonic dystrophy, an unrelated muscle condition. Each patient takes creatine for three months, and at the start and end we make three kinds of measurement: magnetic resonance imaging to measure effects on muscle mass and body composition; a muscle biopsy for laboratory measurements of both the tissue damage by reactive oxygen species and the muscle’s defenses against this; and assessment of effects on muscle strength and the clinical symptoms.

We think this project will be useful in several ways: it will provide evidence of the usefulness of creatine therapy over a longer time span than earlier studies; it should throw light on mechanisms of

muscle damage in FSHD; it may suggest that other substance that reduce oxidative stress in muscle may also be useful as therapy; and lastly, the results will help in the design of future placebo-control trials. We are very grateful for the Society’s support, and look forward to reporting our results in due course.

**Title:** “Muscle damage by reactive oxygen species, muscle atrophy and effects of creatine supplementation in facioscapulohumeral muscular dystrophy.”

**Investigator:** Graham J Kemp, M.D.

**Institution:** University of Liverpool

This project is the first FSH Society Sam E. and Mary F. Roberts Foundation grant for research on FSHD and nutrition. “This is a pilot study designed to test the following hypotheses:

- (1) that muscle in FSHD shows evidence of damage by reactive oxygen species (ROS) in vivo;
- (2) that this is at least due partly to reduced anti-ROS protection;
- (3) that this is ameliorated by six months of creatine treatment;
- (4) that this also partially alleviates muscle atrophy, even in the absence of training; and
- (5) that this results in an increase in muscle strength and clinical indices.

This is an open label pre-post protocol examining the effects of creatine supplementation in 10 patients with proven FSHD (and disease controls and unaffected controls). ROS protection and damage will be studied in conchotome biopsies of biceps. Muscle atrophy and its effect on body composition will be measured by whole body quantitative magnetic resonance imaging (MRI).”

The Society is pleased to have Dr. Kemp and his colleagues at the University of Liverpool, one of the foremost institutions in the world in state-of-the-art MRI and Magnetic Resonance Spectroscopy (MRS), working on FSHD and applying these new methods to examine and quantify critical areas of research.

## Clinical Activity

### FSHD and Genetic Testing Information Available at GeneClinics

A half dozen years ago the Society started working with GeneClinics to help raise the visibility of FSHD. GeneClinics, a non-profit organization, receives funding support from the National Institutes of Health (NIH), the Health Resources and Services Administration, and the US Department of Energy. Technical support for the [www.geneclinics.org](http://www.geneclinics.org) website is by the University of Washington and administrative support is provided by the University of Washington School of Medicine and Children's Hospital Regional Medical Center, both of which are in Seattle, WA. GeneClinics describes its web site on the home page as a "publicly funded medical genetics information resource developed for physicians, other healthcare providers and researchers, at no cost to all interested persons."

GeneClinics asked the Society to help with its last grant application by providing written letters of support for grant applications by the GeneClinics organization to government funding agencies. This led to increased interest in FSHD at the University of Washington and is helping to shore up FSHD research in the northwest and on the west coast of the U.S. For more information, follow the GeneReviews link on the [www.geneclinics.org](http://www.geneclinics.org) website and search for FSHD; this will lead to a summary page on FSHD that contains a section called "Testing;" under "test availability" there is a hyperlink called "testing" that leads to the GeneTest lab search results.

The section will provide details for physicians and genetic counselors on diagnostic and pre-natal testing. The laboratories that conduct clinical testing for FSHD shown on GeneTest-GeneClinics are:

- Alberta Children's Hospital,

Molecular Diagnostic Laboratory,  
Calgary, Alberta, Canada

- Athena Diagnostics, Inc.,  
Reference Lab, Worcester, MA
- Children's Hospital of Eastern Ontario, DNA Diagnostic Laboratory, Ottawa, Ontario, Canada
- Leiden University Medical Center, DNA Diagnostic Lab/KGCL, Leiden, Netherlands
- New York University School of Medicine, Neurogenetics Laboratory, NY, NY
- University Hospital of Umea, Department of Clinical Genetics, Umea, Sweden
- University of Antwerp, Department of Medical Genetics, Antwerp, Belgium
- University of Iowa Health Care, Department of Pathology, Iowa City, IA
- Wolfson Medical Center, Molecular Genetics Laboratory, Holon, Israel

In addition, the Department of Medical Genetics and Neurology, University of Washington, Seattle, WA has posted a web site at <http://depts.washington.edu/neurogen/> called "Genetic Testing for Neurological Conditions" that has informational materials on genetic testing for several diseases. One of the brochures found at this site is called "Facioscapulohumeral Muscular Dystrophy: Making an Informed Choice about Genetic Testing." The FSH Society Scientific Advisory Board (SAB) has not reviewed this document for accuracy. The University of Washington brochure on FSHD is a good read although it has several minor errors. The brochure covers some general concepts in genetic testing and may give the impression that pre-implantation genetic (PGD) diagnosis is available for FSHD. PGD is not yet technically feasible for FSHD according to the top experts in FSHD genetic testing. The FSH Society, through the SAB, continues its work on a brochure for the complex testing aspects of FSHD. The Society hopes to have a definitive and authoritative document for patients and professionals by mid-2003.

### Contributing to the FSH Society is Easy & Convenient

Federal employees and military personnel can donate to the FSH Society, Inc through the Combined Federal Campaign (CFC). Please consider making a contribution to the FSH Society through the CFC. The CFC is operated by the United States Government Office of Personnel Management (OPM). The FSH Society, Inc. CFC code is #2662. For more information about the CFC you may visit the OPM website at <http://www.opm.gov/cfc/index.htm>.

You can also contribute through your United Way program at work.  
Ask your employer to match your contribution.

Remember the FSH Society in your Will.



## Research

### How Does DNA Communicate With Itself? *The FSHD Story*

By Melanie Ehrlich, Ph.D., Professor, Human Genetics and Biochemistry,  
Tulane Medical School and Tulane Cancer Center, New Orleans, Louisiana

Genes are the essential “directors” that code for the proteins that are the main “workers” in every cell of every living organism. Genes are made of DNA. FSHD is a unique disease in the way in which the loss of a very small amount of DNA in chromosome 4 (one of the 23 big packages into which DNA is assembled) indirectly causes the syndrome. It is likely that understanding how this DNA loss in chromosome 4 results in FSHD will not only be of great importance in combating the syndrome, but also in enhancing our general knowledge of the control of turning on and off human genes.

The older, well-established aspect of the control of gene turn-on is that certain proteins “sit” on top of regulatory parts of genes and, that way, make sure, for example, that a muscle-specific gene gets activated in muscle and not in skin. Four of the new and exciting research areas on the mechanisms of controlling human gene turn-on involve the following:

(1) modifying DNA by adding an extra little group to one of its four types of building blocks [called DNA methylation — we have recently learned that abnormal changes in DNA methylation are critical steps in forming most types of human cancers];

(2) modifying special proteins (histones) that are very tightly associated with DNA in chromosomes;

(3) changing the shape of small regions of chromosomes to affect how densely packaged the DNA is in the chromosome or which part of a chromosome touches which other part; and

(4) specifically changing the exact location of part of a chromosome within the compartment (the nucleus) in which it is located.

New research on FSHD in progress in our laboratory and others involves all four of these forefront areas.

Laboratory investigations indicate that one of the ways that FSHD is so unusual is that the loss of a tiny bit of DNA from the very end of chromosome 4 seems to affect turn-on of a gene that must be located on chromosome 4, but quite far away. We know that this very small amount of deleted DNA cannot be responsible for FSHD by itself. Instead, it seems that the loss of this DNA somehow changes communication between two parts of chromosome 4 (long-distance interactions) leading to abnormal activation of a chromosome 4 gene that unfortunately sets in motion the symptoms of FSHD.

Some of the new types of research on the control of gene turn-on involve important interactions of distant parts of chromosomes. Breakthroughs are being made in basic research on unraveling these long-distance chromosome interactions, which are providing needed tools for understanding FSHD. In turn, scientists often learn about new principles of genetics and biochemistry from studying genetic diseases. FSHD promises to yield insights into the workings of long-distance chromosome interactions. We are only beginning to glimpse at this type of DNA-to-DNA communication, which is likely to participate in the regulation of many disease-linked genes in addition to the enigmatic FSHD gene.

[Fern Tsien, Ph.D. and Melanie Ehrlich, Ph.D., Tulane Medical School and Tulane Cancer Center, New Orleans, Louisiana were funded for the period May 1, 2001 to April 30, 2003 under a Marjorie Bronfman Fellowship FSH Society grant (FSHS-MB-006) titled: “DNA Methylation and Chromatin Structure of FSHD-linked Sequences in FSHD Cells, Normal Cells, and Cells from Patients with the ICF Syndrome.” Dr. Ehrlich is continuing the initial work under NIH NINDS R21 grant number NS43974-01 titled: “FSHD Syndrome: DNA Repeats, Methylation, and Chromatin.”]

### Marjorie Bronfman Grant for Molecular Genetics Research on FSHD for 2003

The generosity and commitment of Mrs. Marjorie Bronfman to FSHD research started in 1998. To date, the Society has received six hundred thousand of the seven hundred thousand dollars pledged through 2004. All of the six hundred thousand dollars has been awarded for two-year research fellowships (US\$30,000- US\$35,000/year) for research projects that show extraordinary promise to find the cause of FSHD. This foresighted contribution significantly impacts progress in FSHD research and has already created advances worldwide. The FSH Society is deeply indebted to Mrs. Bronfman and the Marjorie and Gerald Bronfman Foundation for this significant opportunity to advance FSHD research.

### FSHD Patients Needed at the Harvard Neuromuscular Disease Project

By Daniel Paul Perez

The Harvard Neuromuscular Disease Project (HNDP) seeks to understand neuromuscular diseases through research and collaboration. The HNDP is looking for participants and families confirmed to have one of the following neuromuscular disorders: Limb Girdle Muscular Dystrophy, Duchenne/Becker Muscular Dystrophy, Facioscapulohumeral Dystrophy, Myotonic Dystrophy, Miyoshi Myopathy, Myotubular Myopathy, and Centronuclear Myopathy.

Drs. Louis Kunkel and Dr. Robert H. Brown, Jr. are two of the FSH Society’s Scientific Advisory Board members who are key investigators in the project titled “Gene Expression in normal and Diseased Muscle During Development.” The project investigators include: Louis Kunkel, Ph.D.; Robert H. Brown, Jr. M.D., D.Phil.; Alan Beggs, Ph.D.; Emanuela Gussoni, Ph.D.; Elizabeth Engle, M.D.; and Isaac Kohane,

*continued on page 17*

## Research

### FSHD Patients Needed at Harvard Project, *continued from page 16*

M.D., Ph.D.

The HNNDP website at:

[www.tch-genomics.org/hndp](http://www.tch-genomics.org/hndp) has a wonderful overview of the project, its sub-projects, and how you may contribute and become involved. The overview describes "a collaborative effort to use classical methods of gene and protein analysis, as well as state-of-the-art gene expression array technology to study the unanswered questions. Four projects with unique features, but overlapping concepts and methodologies comprise the Neuromuscular Disease Project. The aim of the Project is to identify patterns of gene expression that are global in all dystrophies or distinct to specific sets of dystrophies and myopathies. Ultimately, this will provide insight into the molecular basis of normal muscle development and its dysfunction in these disease states. Long-term, our hope is to use this information in conjunction with the insights from studies of stem cell biology to devise new approaches to the treatment of the muscular dystrophies and related myopathies." Please see internet hyperlink for the entire description:

<http://www.tch-genomics.org/hndp/overview-index.php>

The HNNDP website has a fine

discussion on how to make a difference by becoming a participant and the benefit it may have for the research as well as impact on medical care for individuals and families involved with muscular dystrophies and myopathies. For further information on becoming a participant please see internet hyperlink:

[http://www.tch-genomics.org/hndp/contact-be\\_a\\_partic.php](http://www.tch-genomics.org/hndp/contact-be_a_partic.php)

The HNNDP website also contains a page on Facioscapulohumeral Muscular Dystrophy (FSHD) that is quite informative. Please see internet hyperlink:

[http://www.tch-genomics.org/hndp/pat\\_fam-facio\\_dyst.php](http://www.tch-genomics.org/hndp/pat_fam-facio_dyst.php)

You can contact the Harvard Neuromuscular Disease Project, Harvard Medical School, Children's Hospital in Boston through:

Jessica Blasko, MS, CGC, Research Study Coordinator, Board Certified Genetic Counselor  
Children's Hospital Boston  
Division of Genetics  
300 Longwood Avenue, Enders 5  
Boston, MA 02115.  
617-355-2309, Fax: 617-277-0496

E-mail: [JBlasko@enders.tch.harvard.edu](mailto:JBlasko@enders.tch.harvard.edu),

**Note:** *Tell them the FSH Society sent you!*

### Research Notes

#### FSH Society Announces New Research Fellowship Program

The FSH Society research fellowship program was launched in 1998 under the chairmanship of Dr. David Housman of the Massachusetts Institute of Technology (MIT). In less than five years, we have received numerous inquiries, requested and reviewed letters of intent, and peer-reviewed grant and fellowship applications. We continue to fund and seek out new applications and numerous small projects.

The Society recruits and makes the National Institutes of Health (NIH) aware of researchers expressing interest and data showing scientific promise in FSHD. There are many promising ideas and we are working to assure that each request is given an opportunity for success in funding. We are pushing the leading edge of research and fostering tomorrow's leaders in FSHD research. Since the last newsletter, the FSH Society has funded the following new exciting project:

**Title:** *Restoration of the normal myogenic pattern in FSHD: A nutritional approach.*

Nutritional Grant Roberts' Foundation

**Investigators:** Sara Winokur/Ulla

Bengsston

**Institution:** University of California, Irvine (UCI)

Dr. Winokur has "determined that the process of muscle cell development (myogenesis) is defective in the human disease FSHD. Myoblasts can be monitored in a cell culture system, i.e. growing muscle cell (myoblasts) in the laboratory tissue culture facility. The objective of this study is to identify therapeutic compounds to treat FSHD that can be taken orally as part of a nutritional regimen. Following evaluation of their efficacy in cultured cells, recommendations will be made for potential clinical trials." This would be the first test of any drugs and compounds that act on FSHD cells to see if they affect growth.

### 6th FSH Society Delta Railroad Construction Company Research Fellowship Grant is Established

The FSH Society Delta Railroad Construction Company fellowship program continues to help in the FSHD research efforts by awarding research grants that provide needed expansion of current work and innovative approaches in FSHD studies.

The FSH Society is indebted to the Delta Railroad Construction Company of Ashtabula, Ohio, and Larry and Ida Laurello and their family for this groundbreaking effort on behalf of the FSHD community. The previous five Delta Railroad Research Fellowship Grants are yielding tremendous insights in new and novel areas of FSHD research. We hope that this collaboration will continue and that the members of the Society will consider matching this \$30,000 gift annually.

***The FSH Society is making great strides toward finding a cure. We need your help! Consider donating both financially and with your time!***

## Research

### Second Meeting of the NIH Muscular Dystrophy Research Task Force Held

Daniel P. Perez, FSH Society, participated in the second meeting of the National Institutes of Health Muscular Dystrophy Research Task Force (NIH MDRTF) on "Promoting Treatment," 21-22 January 2003, Bethesda, Maryland along with a dozen other experts in the field of muscular dystrophy. Presentations and topics were:

- Treatments for Muscular Dystrophy, Symptoms being treated, Target Tissues and Organs, Outcome goals, Outcome measures;
- NIH Office of Rare Diseases – Promoting Treatment;
- FDA Office of Orphan Products Development - Orphan Drug Development;
- MDA - Activities Promoting Treatment Development; NIH Translation Research Initiatives;
- Identifying Priority Areas; and
- Promoting cooperation by NIH, Federal agencies, health voluntary groups, and industry.

The Society spoke for the needs of the FSHD community and research. Perez stressed that the design of a platform for launching clinical treatments and trials for dystrophy must be approached with deliberateness and solid scientific discipline, e.g., with structure and references. He asked that, "when embarking on a program for therapy and translational research that it would be desirable to have for each of the nine dystrophies (Duchenne, Becker, limb girdle, congenital, facioscapulohumeral, myotonic, oculopharyngeal, distal, and

Emery-Dreifuss) a summary of the status of current treatments, followed by treatments in the pipeline or expected in the near future. This summary should be followed by an estimate of treatment beyond five years. These can then be used for planning the needed resources to achieve these goals. Alternatively, it might be useful to have nine mini-conferences/research projects to summarize the status of research for each dystrophy and to provide a platform for a thorough review and consideration of unique, distinct as well as non-unique or overlapping aspects of the disorders by the group."

Perez pointed out that several of the dystrophies are simply not at the point to consider gene, vector, cell, stem or small molecule interventions and that much more basic research needs be done vis-à-vis model systems, animal models, mechanisms, phenotype-genotype work. He wants the document to address bringing these dystrophies across this threshold with the needed basic research. This is especially true for Myotonic DM, facioscapulohumeral muscular dystrophy FSHD, and oculopharyngeal OPMD (all diseases of expansion and contraction/deletion of DNA).

When the NIH releases the meeting summary, we will post the complete meeting summary hyperlink from the Scientific Workshops, Conferences, and Committees Reports page on the NIH web site.

The next meeting of the MDRTF will be in late September 2003.

### Current Happenings: Genetic Research at UCLA

Dr. Marahrens and Dr. Embade have contributed the following article to help our readers understand more about their work on FSHD. Please see page 11 for an article from our other new Research Fellow, Dr. Kemp.

#### From the University of California

By York Marahrens, Ph.D., University of California, Los Angeles, United States

Our genes are short segments of our genetic material, the chromosomal DNA molecules. Our chromosomal DNA molecules range from having tens of genes to having thousands of genes. The DNA molecules do not exist as "naked DNA" in the cell. Instead, the DNA molecules are packaged in protein coats called chromatin. Several different types of protein coats (chromatin) exist and different regions of a chromosome are packaged in different protein coats. The various types of chromatin can be placed into two broad categories: euchromatin and heterochromatin.

Euchromatin refers to all of the loose protein coats that allow genes that reside in the euchromatin to be expressed. Heterochromatin refers to all of the tight protein coats that cause genes to be silenced. The tandemly repeated sequences within our chromosomes are packaged in heterochromatin.

Facioscapulohumeral muscular dystrophy (FSHD) arises when patients are missing a number of tandemly repeated DNA sequences that are located near the end of chromosome 4. It is thought that an unidentified disease gene, located somewhere far away from the repeats, malfunctions when the repeats are missing. Since the tandem repeats are packaged in heterochromatin, FSHD patients have smaller regions of heterochromatin near the end of chromosome 4 than healthy people.

Heterochromatin exists at the periphery of the nucleus and euchromatin resides more towards the middle of the

*continued on page 19*

### Second Award for Sam E. and Mary F. Roberts Foundation Grant

The Sam E. and Mary F. Roberts' Foundation of Lawrence, Kansas, established a fund to study nutrition and FSHD. We are indebted to the Board of the Roberts Foundation and its Chairman Susan Pogany for this opportunity.

Drs. Sara Winokur and Ulla Bengsston, University of California, Irvine (UCI), Irvine, California, United States, are the second recipients of the Roberts Foundation grant for their project, "Restoration of the Normal Myogenic Pattern in FSHD: A Nutritional Approach."

## Research

### Dr. Marahrens, Research Fellow, Explains Work With DNA, *continued from page 18*

nucleus. Much of the time, the heterochromatin at the nuclear periphery “sticks” to other heterochromatin. The frequency with which two regions of heterochromatin will stick to each other depends on the locations of the two regions of heterochromatin. There is some evidence that a region of heterochromatin will most frequently form a loop and stick to other heterochromatin that is nearby on the same chromosome. Less frequently, the heterochromatin will stick to heterochromatin far away on the same chromosome. Least frequently, the heterochromatin will stick to heterochromatin located on other chromosomes.

There is also some evidence that a given heterochromatin structure will stick to certain heterochromatin structures but not to other heterochromatin structures. We, therefore, suspect that the heterochromatin structure that exists at the FSHD repeats can only stick to certain heterochromatin structures, and not to other structures.

How might the deletion of the FSHD repeats cause genes to be expressed differently? Genes can be expressed at higher levels if they are located near special sequences called “enhancers of gene expression.” An enhancer located near the FSHD repeats could, in principle, activate a gene that is located very far away if the gene-distant gene is also located near heterochromatin. The gene and the enhancer would be brought near each other when the two heterochromatin regions stick to each other. This sticking would bring the enhancer close enough to the gene to allow it to increase the level of gene expression. If this is true, then the reduction of the size of the region of heterochromatin at the FSHD repeats would cause it to stick to the other heterochromatin less frequently which would cause genes to be expressed at lower levels.

An alternative scenario is as follows. It is also known that genes are silenced if they are located near special sequences

called “repressors of gene expressions.” A repressor, located near the FSHD repeats, could, in principle, silence a gene that is located very far away if the gene-distant gene is also located near heterochromatin. The gene and the repressor would be brought near each other when the two heterochromatin regions stick to each other. If this is happening in FSHD patients, then the reduction of the size of the region of heterochromatin at the FSHD repeats would cause it to stick to the other heterochromatin less frequently and this would cause genes to be expressed at higher levels. We suspect that this second scenario is what is going on in FSHD patients.

We hypothesize that the looping interactions allow the repeats to influence the gene. We are attempting to identify the FSHD genes by attaching a chemical to the repeats in live cells that modifies chromosomal sites that it contacts. The FSHD genes are then identified by virtue of their modification that occurs when they touch the repeats/chemical.

**Title:** *Tethering Adenine (Dam) Methylase to the 3.3-kb FSHD Repeats to Identify Distant Genes that Physically Come in Contact with the Repeats*

**Investigators:** York Marahrens/Neives Embade

**Institution:** University of California, Los Angeles (UCLA)

The Society is honored to know Drs. York Marahrens and Neives Embade and we are delighted to have UCLA working on FSHD. This is a fascinating project that is high risk but has a great potential for profound insight into FSHD. “We propose to locate the FSHD gene(s) that interact with the D4Z4 repeats by tethering bacterial adenine methylase to sequences in or near the 3.3kb repeats and then identifying adenine-methylation at distant sites on the same chromosome or different chromosomes.”

### Listing of Past and Present FSH Society, Inc. Research Fellows

**Researcher:** Silvere M. van der Maarel, Ph.D.

**Institution:** Leiden University Medical Center, Leiden, The Netherlands

**Project Title:** “Generation of Transgenic Mouse Models for FSHD.”

**Researcher:** Sara T. Winokur, Ph.D.

**Institution:** University of California Irvine, Irvine, California

**Project Title:** “Analysis of Chromatin Structure and Skeletal Muscle-Specific Gene

**Researcher:** Denise Figlewicz, Ph.D.

**Institution:** University of Rochester School of Medicine, Rochester, New York

**Project Title:** “Expression of genes proximal to the D4Z4 deletions: a quantitative study in FSHD patients and controls.”

**Researcher:** David J. Picketts, Ph.D.

**Institution:** Ottawa Health Research Institute, Ottawa, Ontario, Canada

**Project Title:** “Utilizing an epigenetic approach to identify the FSHD gene.”

**Researcher:** Davide Gabellini, Ph.D.

**Institution:** University of Massachusetts Medical School, Worcester, Massachusetts

**Project Title:** “Identification and characterization of a protein interacting with the DNA repetitive element causally related to facioscapulohumeral muscular dystrophy.”

**Researcher:** Fern Tsien, Ph.D. / Melanie Ehrlich, Ph.D.

**Institution:** Tulane University Cancer Research Center, New Orleans, Louisiana

**Project Title:** “DNA Methylation and Chromatin Structure of FSHD-linked Sequences in FSHD Cells, Normal Cells, and Cells from Patients with the ICF Syndrome.”

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## Research

### Listing of Past and Present

#### FSH Society, Inc. Research Fellows, *continued from page 19*

**Researcher:** Tonnie Rijkers, Ph.D.

**Institution:** Leiden University Medical Center, Leiden, The Netherlands

**Project Title:** "Mouse models to study candidate genes and epigenetic causes of FSHD."

**Researcher:** Cecilia Ostlund, Ph.D.

**Institution:** Columbia University, New York, New York

**Project Title:** "The role of DUX4 in facioscapulohumeral muscular dystrophy."

**Researcher:** Alexandra Belayew, Ph.D.,  
Stephane Plaisance, Ph.D.

**Institution:** Université de Mons-Hainaut, Mons, Belgium

**Project Title:** "Characterization of a protein expressed from a 3.3 kb element not linked to FSHD" & "Small laboratory equipment for research on FSHD."

**Researcher:** Rossella Tupler, M.D., Ph.D.

**Institution:** University of Massachusetts Medical School, Worcester, Massachusetts

**Project Title:** "Characterization of differentially expressed genes in facioscapulohumeral muscular dystrophy affected muscles."

**Researcher:** Jane Hewitt, Ph.D.

**Institution:** Nottingham University, Nottingham, England

**Project Title:** "Fugu rubripes as a model organism for FSHD gene identification."

**Researcher:** Marcy Speer, Ph.D.

**Institution:** Duke University Medical Center, Durham, North Carolina

**Project Title:** "Genetic Linkage Studies in Non-chromosome 4 FSHD."

**Researcher:** Robert Bloch, Ph.D.

**Institution:** University of Maryland School of Medicine, Baltimore, Maryland

**Project Title:** "Sarcolemmal organization in FSHD and the MYD mouse" & "To investigate the "proteome" in FSHD and to compare it to the "proteome" in control muscles and in other common myopathies and muscular dystrophies using two-dimensional gel electrophoresis"

**Researcher:** Kevin Flanigan, M.D.

**Institution:** University of Utah School of Medicine, Salt Lake City, Utah

**Project Title:** "QMA software/system and professional physical therapy resources to help with studies to answer definitively whether anticipation in disease severity and onset, gender effects, or parent-of origin."

**Researcher:** Jeanne Lawrence, Ph.D./Y.

Polly Xing, M.D., Ph.D.

**Institution:** University of Massachusetts Medical Center, Worcester, Massachusetts

**Project Title:** "Higher level chromatin packaging and nuclear organization of FSHD cell with an emphasis on its 3.3 kb deletion involving high resolution transcript mapping by mRNA in situ and

direct visualization of this region of the chromosome via In situ hybridization with loop halo DNA preparations."

**Researcher:** York Marahrens, Ph.D. /  
Nieves Embade, Ph.D.

**Institution:** University of California Los Angeles, Los Angeles, California

**Project Title:** "Tethering Adenine (Dam) Methylase to the 3.3-kb FSHD Repeats to Identify Distant Genes that Physically Come in Contact with the Repeats"

**Researcher:** Graham J Kemp, M.D.

**Institution:** University of Liverpool School of Medicine, Liverpool, United Kingdom

**Project Title:** "Muscle damage by reactive oxygen species, muscle atrophy and effects of creatine supplementation in facioscapulohumeral muscular dystrophy."

**Researcher:** Sara T. Winokur, Ph.D.

**Institution:** University of California Irvine, Irvine, California

**Project Title:** "Restoration of normal myogenic pattern in FSHD: A nutritional approach."

**Researcher:** Sara T. Winokur, Ph.D.

**Institution:** University of California Irvine, Irvine, California

**Project Title:** "FSHD-Research ListServ."

***The FSH Society has been instrumental in the giant advances in research to find a cure for FSHD. We need your donations to continue the fight!***

***Please see donation form on back page.***

## Community

### FSH Society Groups Welcome New Members and Offers New Resources

Support groups in the Arizona, Colorado, Gulf Area (Alabama, Louisiana and Mississippi), Michigan/Ohio, Mid Atlantic, Minnesota, New England, North West (including Canada) offer the unique opportunity to meet others to discuss Facioscapulohumeral Muscular Dystrophy (FSHD) issues. Meetings are generally held every other month covering topics specific to FSHD. Groups meet in accessible locations. Experts address clinical, research, genetics, nutrition, exercise and coping strategies for FSHD. Individuals, family members and professionals concerned with FSHD are welcome to attend. Information about support groups and networks will be posted on the FSH Society website: [www.fshsociety.org](http://www.fshsociety.org). Please call Karen Johnsen, FSH Society Support Group Coordinator, (301) 262-0701 or email: [kjohnsen5@msn.com](mailto:kjohnsen5@msn.com), with any questions or interest in a local group, telephone network or pen pal group.

We have requests to form groups in San Diego, San Francisco and Los Angeles, CA; Palm Beach, FL; Philadelphia, PA. Please contact Karen for new group information. In order to preserve confidentiality, the FSH Society contacts members directly to inform them of group meetings in their areas.

#### International FSH Society Network Contacts:

- **Australia**

**Ray Jordan**, 86 Barry Street, Reservoir, Victoria 3073, Australia, Phone: 03 9460 2559, email: [rrev@optusnet.com.au](mailto:rrev@optusnet.com.au).

- **Canada**

**Dawn Young**, New FSH Society Network Coordinator for Canada email: [dadayoung@shaw.ca](mailto:dadayoung@shaw.ca)

- **England**

**FSHMD Support Group**, 28 Caldecote Gardens, Bushey, Herts WD23 4GP, England. Phone: 44 20 8950 7500 Fax: 44 20 8950 7300, Web Site: <http://www.btinternet.com/~fsh>, Annual meeting for FSHMD.

- **France**

**Daniel Mennetret**, e-mail: [daniel.mennetret@libertysurf.fr](mailto:daniel.mennetret@libertysurf.fr), liaison to Friends (**Amis**) of FSHD, **Association Française contre les Myopathies (AFM)**, 1 Rue de l' Internationale, 91002 Evry Cedex, France.

- **The Netherlands**

**Dutch FSHD Foundation (Stichting FSHD): Kees C. J. van der Graaf**, President and Founder, **Stichting FSHD**, Kievietslaan 34, 2243 GD Wassenaar, The Netherlands. Phone: 31 70 511 8466 Fax: 31 70 511 0041. The purpose of the foundation is to stimulate, facilitate and fund scientific (genetic and clinical) research into the causes and etiology of FSHD. The Foundation works closely with the FSH Society on scientific issues.

**VSN (Muscular Disease Society Netherlands): VSN FSHD Working Group**, Gortstraat 115, 3905 BD Veenendaal, The Netherlands: **Frank van Zimmeren**, e-mail: [frank\\_van\\_zimmeren@hotmail.com](mailto:frank_van_zimmeren@hotmail.com), liaison with VSN.

- **South Africa**

**Mr. Honiball**, FSHD Coordinator, **Muscular Dystrophy Foundation SA**, PO Box 1535, Pinetown 2123 South Africa, e-mail: [mDSA@megaweb.co.za](mailto:mDSA@megaweb.co.za) **FSH Society's FSHD Chat room in Cyberspace:** Sundays at 2 and 9 p.m. Eastern Time (USA), the Society hosts chats that include our network members worldwide.

#### Additional Resources:

**Videotapes** of selected meetings from the Mid Atlantic FSHD Support Group and New England FSHD Support Group are available on loan. In addition, videotapes of the FSH Society 2002 Network Conference are available. Contact the office of the FSH Society for further details.

**Pen pal network for our teens:** please contact Karen Johnsen, (301) 262-0701 for names of those interested in receiving correspondence.

**Network for the Partners and Family Members:** please contact Dean Johnsen at (301) 262-0701

#### Thank You!

The FSH Society wishes to acknowledge the following for their contributions to our efforts.

Howard and Michele Chabner, CA, for fundraising efforts.

Paul Closson, FL, for hosting the FSH Society Bulletin Board and Chats.

Drs. Melanie Ehrlich, York Marahrens and Graham Kemp for research notes in this newsletter.

FSH Society Members and Donors 2003.

Bill and Judy Herzberg, OR, fundraising for Research and Education Fund and assistance in this area.

Dr. David Housman, Chair, Scientific Advisory Board, for continued dedication to FSHD issues.

Ardeth Millner and Charles Perez, Lexington, MA, for continued support to the FSH Society Office.

Mrs. Betty Schechter for her editorial and writing assistance.

Support Group Leaders: Ann Biggs Williams; Jeri Blom; Lori Heater; Catherine l'Heureux; Linda Hoover; Becky Howell; Karen Johnsen; Meg Morris-Aabakken; Carol Perez; Peg Powers; Joanne Smith; Stephanie Staley; Dawn Young.

Dawn Young, Canada, for starting "The Penny Challenge" and rewarding winners with chocolates.

## Community

### Donations and Contributions

#### Fundraising Events:

**Bill and Judy Herzberg's Friends and Family for the Research and Education Fund 2003:** (see article on front page)

**CA:** Michael Agliardo; Howard Haberman & Martha Lybarger; Anna Marantz; Calvin & Myra Marantz; Scott Marantz; Cathy May Miller; Patrick Nance; Laura Natkins & Peter Gradjansky; Ingrid Rosenthal; Mitchell A. Schoenbrun; Stephen Shotland

**FL:** Norman & Sandra Arky; Sanford & Dorothy Landa; Joan D. Rockhill; Gerald & Pearl Siegel; Morty & Zeta Sudler

**IL:** Karen Hubler

**MA:** Dr. & Mrs. Ronald Arky; Lisa Cohen; John Freedman

**MD:** Cheryl Kollin & Bill Franz; Lori F. Marantz

**ME:** Rubin & Sally Laskoff

**MN:** Christer & Laurel Cederberg

**NJ:** Joseph Bendavid; David Brotman; Louis Goldstein; Alan E. & Harriet Gordon; Mitchell & Marci Heskell; Harold & Edna May Hirshman; Seymour & Lola Kamp; Iris Kislin; Myra & Marvin Marantz; Philip & Susan Marantz; Herman & Martha Rotter

**NY:** Alan Abramson; Anabella & Mark Charwat; Drs. Ruben & Ana Dybner; Albert Fano, M.D.; F. Peter Herzberg, M.D.; Steven & Linda Hill; Elisa & Bill Hirschberg; Maria Katz; Margaret Katz; Michael V. Lanza; David & Elsie Napell; Dr. Leon & Barbara Nesis; Ellen & Alex Passannante; Dr. & Mrs. Hector Retik; Ernesto Seldman, M.D. & Mirta Seldman, M.D.; Paul Zemsky

**OR:** Tracey Barnett & Simon Mvkusic; Susan Bloom & Mac Kieffer; G. Alan & Leslie Comnes; Rebecca Fleischman; Barbara Gilson; Barbara Kim; Mark Saitowitz; Steven Slovic & Rhonda Schwartz; Betty Smith & Peter Korn; Larry Staver & Amy O'Neill; Stacey Vallas & Reinier Warschauer; Courtney & Anthony Vengarick; Milton & Joanne Yatvin

**WA:** Alice & Stephen Goshorn; Vancouver Neurologists, PS

**WI:** Connie & Ernesto Brauer

**Argentina:** Max & Eva Giesen

**Spain:** Anonymous; Andres Giesen; Maximo Giesen



**Friends of Christopher Stenmon - Fifth Annual End of Tax Season Events in Quincy, MA** to support the activities of the FSH Society 2003:

Alba Bar and Grill; Gloria Andersen; Lara Apovian; Bad Abbots; Megan Billingham; John Bryson; Sheri Buckingham, Jr.; Thomas Carlo; Stephen Cavicchi; Club 58; Bonnie Colombo; Crosshaven Partners; Chad Dagraca; Kevin R. Danehy; Darby's Public House; Erin L. Delaney; David DiIulis; Kimberly Eddy; Eight Ball Billiard Parlor; Inc.; Andrew W. Fink; Jason Forish; Carrie Hartman; David R Huck; Dawn G Kurtz; Le Disco, Inc.; Scott Lennon; Francis P. Leone; Joanne P. Maguire; Joseph Marnikovic; Jennifer Maya-Salamone; Anne McDonnell; Mary-Jeannette Alderman McGarry; Carol Murphy; O'Connor & Drew; P.C.; Mary Oldmixon; Kimberly A. Ollerhead; Patrick K. Faherty, Law; Mary F Ray; Deirdre J. Roach; Elena C Roberts; Michelle J. Rota; Michael A. Rozman, NY; Sarsfield, Inc.; Jennifer Spadorcia; Christopher Spillane, NH; Geraldine Spillane; Russell C. Stamm; Julie Steinkrauss; Carol & William Stenmon; Christopher Stenmon; Frances A. Uftring; Dianne Wolpert; Liqin Zhu; Sharon Zidek (All donors located in MA except where noted).



**High Woods Sportsman's Club Fourth Annual (2003) FSH Society Benefit Archery Shoot**, Saugerties NY, Proceeds from the archery shoots were donated to the FSH Society. With special thanks to **John and Denise Van Etten** for their good work on our behalf.



#### Foundations

Alliance Insurance Charity Program MN; Anheuser Busch Matching Grants Program MO; Anonymous Donor, The New York Community Trust Foundation NY; Mrs. Marjorie Bronfman, Marjorie and Gerald Bronfman Foundation Montreal, Canada; Delta RR Construction Company OH; Gerald Norton Memorial Foundation IL; Ephraim and Karen Heller

Philanthropic Fund CA; The J.P. Morgan Chase Foundation; Sam E. and Mary F. Roberts Foundation KS



#### Corporate Matching Funds

Crosshaven Partners MA; Merck Matching Funds; National Football League NY; Research Systems, Inc. CO



#### United Way

IBM; Price Waterhouse; Schering Plough; United Way of Central Maryland



*The following individuals requested to be acknowledged in the FSH WATCH as of June 15, 2003:*

#### New & renewing members

Anita McCoy OR; Sanford L. Batkin NY; Steven Blier NY; Jerilyn Blom MN; Lori Calandro MI; Gary Cohen, M.D. NY; Mr. Charles M. Fitts, Jr. MS; Dr. & Mrs. G. Isaacson NJ; Susan Jantrzen KS; Robert Kobrin CA; Daniel & Ruth Krasner NY; Doris Olds Eck MD; Linda Passon MN; Judith and Allan Rosenblum IL; Robert Trumble MI



#### Professional Member

William Herzberg, M.D. Portland, OR; \*Neurologist with Special Interest in FSHD



#### Donor

Morty & Zeta Sudler FL



#### In Honor of:

- Peggy Browning; Susan & Stefano Pogony KS
  - Joyce & Bernard Chabner's 50th Anniversary: William & Sylvia Shapiro IL; Dr. & Mrs. Seymour B. Siegel IL
  - Howard & Michele Chabner:
- CA:** Daniel F. Cooley; Kathy Dees & Dwight Dickerson; Howard & Susu Fine;

*continued on page 23*



## Community

### In Honor of, *continued from page 22*

Doug Norton & Paula Jackson; Mannan & Margaret Latif; Mary Savoy; Grace Shohet & David Brownstein; Joey Thyret; Roz & Judd Wenner

**GA:** M. Snyderman & V. Dunnigan

**IL:** Michael & Marla Craven; Allen & Julie Glass; Stewart L. & Rochelle Grill; Betty & Sherwin Korey; Marsha & Jerry Kraut; Mark & Wendy Perlman; Michael & Lisa Radin; Judy & Allan Rosenblum; Paul & Shirley Rustin; Aaron & Suzie Siegel; Dr. & Mrs. Seymour B. Siegel; Stephanie & Michael Smerling

**MA:** Mike & Lisa Heyison

**TX:** Elliott & Sharon Slusky

**VA:** Ronald & Susan Stern

- Justin Zachary Cohen on his Twelfth Birthday: Harriet & Stuart Cohen NY; Judith Cotler NY; Rose Kanter NY

- Sarah Love Davis: Mr. Charles M. Fitts Jr. MS

- Mr. & Mrs. Mark Devore on the arrival of their son, Joshua: Elaine Disick MA

- The Leonard Gilman Family: Elaine Disick MA

- Mr. & Mrs. Leonard Gilman 50th Anniversary: Joyce & Robin Blatt & their families; Myra Blatt; Jodi & Kenneth Farber

- Miriam Kaplan's 80th Birthday: Rose Kanter NY

- Kaaren Lobel's Special Birthday: Carol & Paul Cohen; Diane L. Hockstein; Nathan & Ethel Neibauer; Jeff & Mary Schwartz; Karen Strauss

- William G. Michael, "A Gentleman and a Scholar": Henry Wiggan MA

- Myra Marantz's 70th Birthday: Stanley Arky

- Nicholas Pogany:

Grandmother; Mrs. Clara Pogany

- Mr. & Mrs. Joseph Polonsky's 50th Anniversary: Mr. & Mrs. Stuart Cohen FL

- Mr. & Mrs. Irving Rappaport's Anniversary: Rose Kanter NY; Joseph & Miriam Kaplan NY

- Jessica Ryley: Mary Doto NY; Rick & Leslie Frye WA; Joanne & Gerry Smith MI; Timothy, Cassie & Matthew Smith TX

- Marc Schindelheim's 50th Birthday: Gary Cohen, M.D. NY; Harriet & Stuart Cohen NY; Rose Kanter NY;

Caryn Stoner NJ

- Samantha Schindelheim's Bat Mitzvah: Judith Cotler NY

- Steven Sicker's 60th Birthday:

Stuart Cohen; Eli Schindelheim

- Helen Younger: Jane Batkin CT



### In Memory of:

- Laura Barsh,

**NJ:** Robert & Marlene Krum; National Football League; Amy Abromson; Sandra Alken; Ellen Barlow; Megan Fenerty; Chris Hangle; Cindie Hurley; Lorena Pannizzo; Marie Patriarca; Erica Pitt; Damon Reynolds; Jenn Rodia; Chrissy Shaughnessy; Kari Shisler; Lisa Silva; Colleen Smith-Grubb; Brian Song

**PA:** Bill & Ellen Alesker; Deborah Angler; Flora & Herbert Berk; Ted Berk & Family; David & Charlotte Bernstein; Dr. & Mrs. Franklin Brody; Helen Brookland; Dr. & Mrs. Malcolm Ecker; Mr. & Mrs. Stanton Friedman; Henrietta Goldfarb; Barbara Goldstein; Barbara Gordon; Patricia Gevurtz; Dr. & Mrs. Marvin Kanefield; Sharon & Saul Katz; Harold & Roslyn Leese; Maribeth & Steven Lerner; Penny Orloff; Michael & Martha Richino; Binnie Sharp; William & Barbara Shoulberg; Ruth & Alan Smith; Debra Lee Steinberg; Jill A. Talus; Gary & Ann Udis; Irwin & Marcy Ufberg; Cheryl Weiss

- Claudine Brown: Daughter; Dr.

Myrna Dingman NM

- Lorna Everett: W.D. Jr. & Judith S. Ross WA

- Stanley Futterman: Mr. & Mrs. Eli Schindelheim FL

- Debra Glann: Larry & Ida Laurello
- Sam Goldstein: Rose Kanter NY
- James Healy (New York):

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**NY:** Mr. & Mrs. B. Alessi; Mr. & Mrs. S. Anderson; D Glenn & Linda Bernhard; Mr. & Mrs. Harvey Bloom; John & Regina Buckley; Joseph & Nancy Byrne; Mr. Michael Cinquemani; Mrs. Rosalie Cinquemani & Family; Jean Coghlan; Christopher & Eileen Connolly; Denyse De Stefani; Thomas & Doreen Dorsa ; Dorsa; Courchesne & Laudage Families; Mr. & Mrs. J. Fiorello; Ronald Fregara; Mrs. Carol Fregara & Family; Pat Galinkas; Mr. & Mrs. D. Gish; Donna Lane; Art Wisnieski & friends at Lockheed Martin; Mr. & Mrs. G. Mehr; Richard & Dolores Muller; Mr. & Mrs. V. Pisciotta; Pat Powell; Donna Slattery; Mark Stavin; Mr. & Mrs. H. Sylvan; Stephen Valdini; Mr. & Mrs. K. Walsh; Mr. & Mrs. J. Woodstock; Nina T. Zash

- Barbara Peterson: W.D. Jr. & Judith S. Ross WA

- Fred Shirk: Robert & Dolores Martin CA

- Mr. & Mrs. Homer Smith: W.D. Jr. & Judith S. Ross WA

- Burt Stone: Judith Cotler NY; Eli Schindelheim NY

- John A. Stout: Ted & Meg Klein OH

- Margaret (Peggy) Whitehurst: Eric & Michelle Breiding; Dean & Stacy Dockus; Janice M. Greavu; Jean Nuss; Mary Redle; Pat Russ; John & Marge Seitz

If you know someone who . . .

would be interested in knowing about the FSH Society and our work

or someone who would like to support our efforts

or perhaps a physician who should be aware of FSHD . . .

please call the east coast office at (781) 860-0501, give us their name and address and we will be glad to send them the newsletter and other

information about FSHD.

# FSH Society Membership Application—Donation

Name(s) \_\_\_\_\_

Address \_\_\_\_\_

City/State/Zip \_\_\_\_\_

Phone: Day (    ) \_\_\_\_\_ Evening (    ) \_\_\_\_\_ E-mail: \_\_\_\_\_

Use space below for address changes, specifying interested individual, specifying title and affiliation, corporate designation or for any other comments you may have:

\_\_\_\_\_  
\_\_\_\_\_  
\_\_\_\_\_

## Individual Membership

- Regular Member \$35.00 +
- Sustaining Member \$70.00 +
- Patron \$100.00 +
- Sponsor \$250.00 +
- Donor \$500.00 +
- Benefactor \$1000.00 +
- Other \$ \_\_\_\_\_

## Professional Membership

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- Corporate I \$1000.00 +
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## Non Membership Donation

- Other \$ \_\_\_\_\_

## Research & Education Fund Donation

- Other \$ \_\_\_\_\_

Total enclosed \$ \_\_\_\_\_

- Please check here if you would like your membership or donation acknowledged in the next issue of the FSH newsletter.

*Contributions to the FSH Society are tax-deductible and acknowledged for tax purposes.*

*Please make checks payable to the FSH Society and send contributions to:*

*Carol A. Perez, Executive Director, FSH Society, Inc. 3 Westwood Road, Lexington MA 02420 USA*

***Please Note: Checks or money orders from outside the United States should be in US dollars from institutions with US bank affiliations.***