Testimony of Daniel Paul Perez, President & CEO of the FSH Society before the Subcommittee on Labor, Health and Human Services, Education and Related Agencies on the Subject of FY2009 Appropriations for National Institutes of Health (NIH) Research on Facioscapulohumeral Muscular Dystrophy (FSHD)
March 20, 2008

Mr. Chairman, thank you for the opportunity to testify.

I am here to remind you that muscular dystrophy (MD) is still taking its toll. As a patient with facioscapulohumeral muscular dystrophy (FSHD), I have experienced the constant loss of function this disease leaves in its wake.

We request this year in FY2009, help for those of us coping with and dying from MD and FSHD, as we did in FY2008. **Specifically we ask that Congress and the Appropriations Subcommittee on Labor, HHS, Education and Related Agencies consider:**

1. Resuming the five year doubling of the National Institutes of Health (NIH) budget. Over the past year the research funding situation has gone from bad to worse and we have lost opportunities to fund excellent research.

2. Appropriating $80 million to MD research at the NIH in FY2009 and steadily increasing this amount to at least $125 million annually over the next five years.

3. Making NIH funding comprehensive for basic research in each of the nine types of MD as well as creating an equitable distribution for each MD across the Senator Paul Wellstone Cooperative Research Centers network, program projects, clinical research, and translational research programs.

Last year, the Honorable Chairman Rep. David Obey asked us to go back to the districts and tell fellow citizens that many of these programs cannot be funded due to the current tax cuts. Five years prior, I testified before this same committee and the exact same thing was said. Given the extent of my physical disability and the Herculean effort it takes me to be present, I personally found this advice very painful. It is irresponsible of Congress to ask those of us who are sick and dying to compensate for the actions of the Congress and the Administration. It simply is not right for Congress to put the burden back on the shoulders of those with disability, disease and little means to effect decisions made by the Congress. Let me be on record that the Boston Globe did an interview with us and ran a story on our testimony and on Mr. Obey's request to us to help get his message out. We have done what you requested of us.

Our first request calls for increasing the NIH budget and resuming the five year doubling. The wars in Iraq, Afghanistan, tax cuts and the turmoil in the financial markets have essentially halted any progress in biomedical research. We all know that America has fallen far behind in biomedical research funding. As a person with a disease it is hard to reconcile the generosity of the Congress towards the wars, tax cuts to the wealthy, and bailing out institutions that have put us all at financial risk, against the lack of action on behalf of sick and dying citizens. The NIH budget at $29.2 billion is a miniscule fraction of these other expenses. Doubling a tiny fraction is still a tiny fraction. For those in Congress who ask the NIH where are the cures – consider that the NIH budget of $29.2 billion covering countless thousands and thousands of diseases is a fraction of the market capitalization of a large pharmaceutical company covering a few disease areas. Consider also that the main job of NIH is basic science, not drug development, and that the pharmaceutical companies, the American public and people

throughout the world benefit directly from the NIH investment in science. Please act now to refocus spending on American infrastructure before trust and confidence is lost.

Our second request calls for NIH to build and grow its MD disease area funding to a level commensurate with diseases of similar burden. A wide disparity still exists in funding for MD. This is a matter for both Appropriations and for the NIH, with its wide discretion on funding for diseases. More funding would help balance out these disparities and accelerate treatments and cures for diseases. We request that the Director of the NIH consider a more commensurate and equitable amount for MD that is solidly in line with its disease peers at $80 to $125 million.

Our third request asks the Appropriations Committee to request that the Director of NIH increase the amount of FSHD research and projects in its portfolios using all available mechanisms and interagency committees. Given the knowledge base and current opportunity for breakthroughs in treating FSHD it is inequitable that only two of the twelve NIH institutes covering muscular dystrophy have a handful of research grants for FSHD and that FSHD research is virtually non-existent in the Senator Paul Wellstone MD Cooperative Research Centers (CRCs). This funding should include projects from NIH roadmap, extramural programs, intramural programs, Senator Paul Wellstone MD CRCs and similar program projects that have a major focus on FSHD.

FSHD is the second most prevalent adult muscular dystrophy. The incidence of the disease is conservatively estimated to be 1 in 20,000. The prevalence of the disease, those living with the disease ranges from 15,000 to 40,000 Americans based on our increasing experiences with the disease and accurate diagnostic tests. For men, women, and children the major consequence of inheriting FSHD is a lifelong progressive and severe loss of all skeletal muscles. FSHD is a terrible, crippling and life shortening disease. It is genetically transmitted to children and it affects entire family constellations.

The Appropriations Committee is no stranger to asking the National Institutes of Health (NIH) for more research on facioscapulohumeral muscular dystrophy (FSHD). More than eight years ago, on February 15, 2000 the House Appropriations Committee questioned Dr. Ruth Kirschstein, then the Director of NIH, on why FSHD research has been slow to develop. In addition, Dr. Gerald Fischbach, then the Director of the National Institute of Neurological Disorders and Stroke (NINDS), was asked to respond to questions about the NIH non-response to developing FSHD on February 29, 2000. This inquiry followed a period of five or six years of miniscule growth in FSHD research portfolios at the NIH.

In late 2000, we shifted our efforts to the authorization track. It was an honor, then, to help the late Senator Paul Wellstone rewrite portions of the Muscular Dystrophy Community Assistance, Research and Education Amendments of 2001 (the MD-CARE Act, Public Law 107-84) to include all nine types of muscular dystrophy, including women, adults and not just children with Duchenne muscular dystrophy (DMD). We also added patient representative and input at the inter agency level via the Muscular Dystrophy Coordinating Committee (MDCC) advisory committee. The MD-CARE Act was passed mandating the NIH and other applicable federal agencies to immediately expand and intensify research on each form of muscular dystrophy.

How is facioscapulohumeral muscular dystrophy (FSHD) research at NIH doing in 2008, seven years after the MD CARE Act 2001 was passed, and, fifteen years after our first testimony in person before the late Representative Natcher, a former Chairman of this honorable Committee?

We applaud Dr. Story Landis, Director, National Institute of Neurological Disorders and Stroke (NINDS), and, Chairman of the MDCC; Dr. Stephen Katz, Director, National Institute of Arthritis and Musculoskeletal Disorders (NIAMS) and past-Chairman of the MDCC; Dr. John Porter, Program Director Muscular Dystrophy, NINDS and Executive Secretary of the MDCC, and; Dr. Glen Nuckolls, Program Director Muscular Dystrophy, NIAMS for their extraordinary comprehension, insight, accuracy and speed with which the NIH Action Plan for Muscular Dystrophy was researched, compiled, written, and approved. The NIH is making significant investments to understand muscular dystrophy research needs and has made excellent choices in recruiting program staff with the ability to understand the extremely complex nature of MD.

Between fiscal year 2006 and 2007, NIH overall funding for muscular dystrophy increased from $39,913,000 to $47,179,000, an 18 percent increase. Figures from the NIH Appropriations History for Muscular Dystrophy show that from the inception of the MD CARE Act funding has doubled for muscular dystrophy.

Between fiscal year 2006 and 2007, NIH funding for facioscapulohumeral muscular dystrophy (FSHD) increased from $1,732,655 to 4,108,555. In fiscal 2007, FSHD was 8.7% of the total muscular dystrophy funding ($4.109M / $47.179M).

National Institutes of Health (NIH) Appropriations History
Source: NIH/OD Budget Office & NIH OCPL (Dollars in millions)

Fiscal Year	NIH Overall Dollars	MD Research Dollars	FSHD Research Dollars	FSHD % of MD
2000	$17,821	$12.6	$0.4	3%
2001	$20,458	$21.0	$0.5	2%
2002	$23,296	$27.6	$1.3	5%
2003	$27,067	$39.1	$1.5	4%
2004	$27,887	$38.7	$2.2	6%
2005	$28,494	$39.5	$2.0	5%
2006	$28,587	$39.913	$1.7	4%
2007	$28,899	$47,179	$4.109	8.7%
2008	$29,230Est	$47,221Est	-	-

FSHD is the second most prevalent adult muscular dystrophy after myotonic muscular dystrophy (DM). We are very concerned about the wide disparity in funding between the most widely recognized pediatric muscular dystrophy (DMD) and the entire group of the other eight types of MD. DMD has exclusive funding from the Centers for Disease Control (CDC), Department of Defense (DoD) and more than half (>50%) of NIH funding for MD. This is astounding considering FSHD and DM are each more prevalent than DMD and each received 5% and 15% respectively of total MD dollars as last reported by the NIH to Congress!

Between 2006 and 2007, the NINDS became the lead institute for funding in MD. Historically, the NIAMS in its mission statement has been primarily responsible for and has been the lead institute for muscle disease research. The Center for Scientific Review (CSR) routes the majority of MD grant applications to NIAMS based on its mission. In fiscal year 2007, NIAMS was the second largest contributor, followed by NICHD as third, and NHLBI as fourth. It should be concerning that muscular dystrophy spending has declined significantly in several key institutes that could bring tremendous impact to these devastating diseases.

National Institutes of Health (NIH) Muscular Dystrophy Funding by Institute FY2007
Source: NIH/OD Budget Office (Dollars in millions)

Participating ICs	FY 2006 Actual	FY 2007 Actual	Change
NINDS	12.697	19.347	+51.6%
NIAMS	16.576	17.734	+7%
NICHD	4.818	4.591	-4.7%
NHLBI	2.270	2.458	+8.3%
NIA	1.865	1.882	+0.9%
NCRR	0.770	0.679	-11.8%
NCI	0.495	0.426	-13.9%
NHGRI	0.391	0.161	-58.8%
NINR	0.031	-	-
NEI, NIMH, FIC, OD	0.0	0.0	0%

NINDS: In fy2007, NINDS spent $2,612,994 on FSHD and $19,247,940 on MD. 47 projects, including Wellstone CRC components for a total of $19,247,940 were funded. FSHD was 13.6 percent of NINDS MD funding. The NINDS funded, for FSHD, three research grants, one research fellowship, one research contract, one-quarter of a Wellstone MDCRC center and one-half of a Translational Research Center research grant for a total of six projects. NINDS funding for FSHD went up by $1,191,398 or 83.8%. Total funding for MD by NINDS increased over the year by $6,551,266 or 51.6%.

NIAMS: In fy2007, NIAMS spent $1,495,561 on FSHD and $17,734,317 on MD. This comprises 89 projects, including Wellstone MDCRC components for a total of $17,734,317. FSHD was 8.4 percent of NIAMS MD funding. The NIAMS funded, for FSHD, four research grants, one research contract, 2% of a Translational Research Center for a total of six projects. NIAMS funding for FSHD went up by $1,184,502 or 381%. Total funding for MD by NIAMS increased over the year by $1,158,000 or 7%.

NICHD: In fy2007, NICHD spent $0 on FSHD and of $4,591,826 on all MD. 17 projects, including three Wellstone MDCRC centers were funded. FSHD was zero percent of NICHD MD. Total funding for FSHD by NICHD was $0. Total funding for MD by NICHD decreased over the previous year by $225,756 or 4.7%.

NHLBI: In fy2007, NHLBI spent $0 on FSHD and $2.458 million on MD. FSHD was zero percent of NHLBI fy2007 MD funding. Total funding for FSHD by NHLBI remained at zero dollars. This should be of grave concern as respiratory insufficiency and failure is becoming increasingly recognized as a cause of poor quality of life and, even more significantly, of death in FSHD.

The MD CARE Act 2001 mandates the Director to intensify efforts and research in the muscular dystrophies, including FSHD, across the entire NIH. It should be very concerning that only two of the institutes at the NIH are funding FSHD. NICHD, NHLBI, NHGRI, NCI and NCRR are all aware of the high impact each could have on FSHD. FSHD is certainly still far behind when we look at the breadth of research coverage NIH-wide.

Centers of excellence -- Wellstone MD Cooperative Research Centers (MDCRCs, U54s) are mandated by the MD CARE Act and, to date, have not been established to cover each of the nine types of MD. There is an inequitable distribution of each of the muscular dystrophies across the Wellstone centers with almost two-thirds of the entire center network, four out of six centers, focusing on DMD. FSHD has about a five (5) percent share of the entire current Wellstone centers

portfolio. Today, the NIH has six Wellstone centers, but they have almost no presence for FSHD, and nothing at all for related dystrophies such as Emery-Dreifuss Muscular Dystrophy (EDMD) and Oculopharyngeal muscular dystrophy (OPMD).

I am here once again to remind you that FSHD is taking its toll on your citizens. FSHD illustrates the disparity in funding across the muscular dystrophies and recalcitrance in growth over twenty years despite consistent pressure from appropriations language and Appropriations Committee questions, and an authorization from Congress mandating research on FSHD.

We request that Congress resume the doubling of NIH funding every five years. Under the current budget, research funding percentiles have reached the top tenth percentile and higher. With funding pay lines at ten percent, plus or minus a few percent, excellent research proposals are going unfunded. We request that the Appropriations Committee to act now to restore the lifeline to biomedical research in the United States to avoid an accelerated loss of researchers and clinicians.

We request that $80 million to $125 million annually be appropriated for MD. We all know that for a disease area to grow -- grant applications must be received and grant applications must be funded. The majority of growth in any disease area at the NIH is obtained through unsolicited applications. In the widest sense, in order for NIH to increase the MD portfolio across the missions of applicable and participating institutes more funding is needed for the NIH. We request the Appropriations Committee help increase the number of unsolicited FSHD and MD grants awarded by lowering the pay lines with an increase in the overall pool of funds NIH works with.

We have learned from experience that the FSH Society as a volunteer health agency and the patients it represents serve a vital function in developing research. We develop an area of research to a point where the NIH could then push the research to much greater heights. The FSH Society has invested over $2 million in the last nine years into nearly seventy post-doctoral and research fellowships and grants. In the last two years, our understanding of how FSHD mechanistically works has dramatically increased. This, in turn, allows researchers to fill the gaps between mechanistic knowledge to translational research to clinical trials. This knowledge has dramatically increased thanks to the efforts of patients, the FSH Society, researchers, clinicians and the NIH. Investments from small non-profits like the FSH Society have allowed the global funding and initiation of novel challenging and promising research in FSHD. Two of the three research projects funded by NINDS are past FSH Society research fellows (5-R01-NS048859-04 M. Ehrlich, 5-R01-NS047584-05 R.G. Tupler). Three of the four research projects funded by NIAMS are past FSH Society research fellows (1-R01-AR-52027-01-A2 Y.W. Chen, 1-R01-AR-56129-01-A2 R.G. Tupler, 1-R21-AR-55876-01 S. van der Maarel) and the fourth project came from FSH Society patient networking activities (1-R01-AR-55877-01 P.L. Jones).

We request that the Director of the NIH be more proactive in facilitating grant applications (unsolicited and solicited) from new and existing investigators and through new and existing mechanisms, special initiatives, training grants and workshops – to bring knowledge of FSHD to the next level.

Thanks to your efforts and the efforts of your Committee, Mr. Chairman, the Congress, the NIH and the FSH Society are all working to promote progress in facioscapulohumeral muscular dystrophy. We are pleased to see FSHD funding from the NIH and federal research agencies gaining traction. FSHD funding is just now beginning to grow. Our successes are just beginning and your support must continue and increase.

We ask you to fund biomedical research, fund NIH, fund MD, and fund FSHD.

Mr. Chairman, thank you for this opportunity to testify before your committee.