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 FY2008 Appropriations for National Institutes of Health (NIH)
Research on FSH Muscular Dystrophy (FSHD)
March 27, 2007

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

I know you are busy with the wars in Iraq, Afghanistan and tax cuts. I am here to remind you that facioscapulohumeral muscular dystrophy (FSHD) is still taking its toll on your citizens.

I am no stranger to this process and FSHD. 14 years ago, I presented to Representative Natcher and have sat at this table many times since.

FSHD is the second most common dystrophy, a crippling disease causing loss of all skeletal muscle, and it affects 20,000 Americans.

The unique epigenetic structure of FSHD is unprecedented in other genetic disorders and it demands novel approaches and new research groups. There are no free lunches. Investments from small non-profits like the FSH Society and NIH have allowed the initiation of novel challenging and promising research. Our successes are just the beginning, we are not there yet and your support has to be continued. It is vital to follow multiple and complementary approaches.

Today, I ask you for three things.

1. Resume the five year doubling of the NIH budget. Appropriate $32.8 billion as required in the NIH Health Reform Act 2006 (P.L. 109-482). At minimum, appropriate a 6.7% annual increase over the next three years to restore funding. Only America has the ability to solve devastating dystrophies. Only NIH can do this. The question is -- does America have the will? Small non-profits like the FSH Society can not shoulder the burden year after year for new and novel research funding. This is what NIH is designed to do. Congress needs to fund it.

2. Tell the Director of NIH to make the Muscular Dystrophy Research Action plan submitted to Congress viable. The MD CARE ACT 2001 authorization needs an appropriation with it. It requires $100 million in FY2008 and $250 million over the next five years for NIH. This will stop dystrophy dead in its tracks and return on investment will be immediate. Tell the Director that the "Pioneer" and "Roadmap" programs have taken money away from peoples' diseases. Tell him to spend the "Common Fund" directly on research that will effect patients' lives.

3. Tell the Director that $1.7 million for FSHD does not cut it. The funding for dystrophy is not equitable. The funding for FSHD is abysmal. FSHD research will help solve others diseases such as cancer, autism and diabetes. Tell the Director to assign $20 million for FSHD.

Congress has repeatedly asked NIH for a comprehensive portfolio on FSHD. I wrote portions of the MD CARE ACT 2001 with Senator Wellstone. I have served for six years on the Muscular Dystrophy Coordinating Committee (MDCC). I have done everything that the Administration and NIH has asked of me. I implore you to ask the Director why, after all this time and effort, only one institute has funded research grants for FSHD?

The war will end and tax cuts will be resolved in 2008. You know, I know and the American public knows that America has fallen behind when it comes to health care and biomedical research funding.

Fourteen years ago, I was able to walk into this room. Today, I appear before you in a wheelchair.

We are waiting.
While we wait, people like me are losing our battles with FSHD.
While we wait, we are losing our quality of life.
Research must be funded and hope rekindled.
I ask you to fund NIH, fund muscular dystrophy, fund FSHD.
I will answer any questions.

Facts on FSHD and NIH funding on FSHD

Facioscapulohumeral Muscular Dystrophy

FSHD is the second most prevalent adult muscular dystrophy. It affects 20,000 Americans. For men, women, and children the major consequence of inheriting FSHD is a lifelong progressive and severe loss of all skeletal muscles. FSHD is simply a most terrible and crippling disease. It is genetically transmitted to children so it affects entire family constellations.

People who have FSHD must cope with continuing, unrelenting, unpredictable 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. But 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.

Insidiously and systematically, 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, and the way people perceive 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.

The MD-CARE Act, P.L. 107-84 and the NIH

After ten years of our testifying, begging and pleading for help, the Congress enacted the **Muscular Dystrophy Community Assistance, Research and Education Amendments of 2001** (the MD-CARE Act, Public Law 107-84). Both the Senate and House acted with force and clarity to mandate the NIH and other applicable federal agencies, to immediately expand and intensify research on all forms of muscular dystrophy. The MD-CARE Act declared that: 1.) the Director of the NIH work with the Directors of the National Institute of Arthritis and Musculoskeletal Disorders (NIAMS), the National Institute of Neurological Disorders and Stroke (NINDS) and the National Institute of Child Health and Human Development (NICHD) and others to expand and intensify research on all nine types of dystrophy described in the Act; 2.) Centers of excellence for research be established for all nine types of dystrophy; 3.) a MDCC with two-thirds government and one-third public members be established to coordinate activities across the NIH and other national research agencies on all forms of dystrophy; and; 4.) the MDCC must submit a research action plan for

conducting, and supporting research and education for all nine types of dystrophy. The MD-CARE Act also requires annual updates on research funding amounts by the Department of Health and Human Services (DHHS) for Duchenne, Myotonic, FSHD and other muscular dystrophies.

How is FSHD Doing in 2007, Six Years After the MD CARE Act was Passed?

The Act mandates the Director to intensify efforts and research in the muscular dystrophies across the NIH. However, all nine types of dystrophy have not benefited and FSHD is certainly falling far behind.

Centers of excellence otherwise instituted as cooperative research centers (U54s) have not been established for all nine types of muscular dystrophy. FSHD has very little presence in the current Wellstone MD Cooperative Research Centers landscape. The NIH now has six Wellstone MD CRCs, which are approximately equivalent to 31 basic research grants (R01). One-quarter of one Wellstone MD CRC, and two-fifths of another Wellstone MD CRC or three R01 grant equivalents have direct relevance to FSHD.

We applaud Dr. Stephen Katz, Director, NIAMS and Chairman of the MDCC, and John Porter, Program Director Muscular Dystrophy, NINDS and Executive Secretary MDCC, for their extraordinary comprehension, accuracy and for the speed in 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 muscular dystrophy. The plan was submitted two and a half years ago and many of the items require lead time and require increased funding. However, to this day, the NIH reports difficulty in growing and expanding its FSH muscular dystrophy research portfolio.

National Institutes of Health (NIH) Muscular Dystrophy Dollars by Institute FY2006
Source: NIH/OD Budget Office (Dollars in millions)

Participating ICs	FY 2006 Actual
NIAMS	16.6
NINDS	12.7
NICHD	4.8
NHLBI	2.3
NIA	1.9
NCRR	0.8
NCI	0.5
NHGRI	0.4
NEI, NIMH, FIC, OD	0.0

NIAMS

In fy2006, NIAMS spent $311,059 on FSHD and $16,575,863 on all muscular dystrophy. This comprises 74 projects, including three Wellstone MD CR centers. FSHD was 1.9 percent of NIAMS dystrophy funding.

No research grants, one research contract for FSHD, the National Registry for myotonic and facioscapulohumeral muscular dystrophy for $310,559 and one conference grant for $500.

NIAMS funding for facioscapulohumeral muscular dystrophy went up by $15,171.

Total funding for muscular dystrophy by NIAMS decreased over the year by 3.3%.

This should be shocking to anyone reading this report; NIAMS is the leading institute for muscular dystrophy at the NIH.

NINDS

In fy2006, NINDS spent $1,421,596 on FSHD and of $12,696,674 on all dystrophy. 39 projects, including two Wellstone CR centers were funded. FSHD was 11.1 percent of NINDS muscular dystrophy funding.

Three research grants, one intramural grant, and one-quarter of a Wellstone CR center for facioscapulohumeral muscular dystrophy.

NINDS funding for facioscapulohumeral muscular dystrophy went up by $61,666.

Total funding for muscular dystrophy by NINDS increased over the year by 6%.

The FSHD boat is sinking in the rising tide, concerning to anyone reading this report.

NICHD

In fy2006 NICHD spent $0 on FSHD and of $4,817,582 on all dystrophy. 19 projects, including three Wellstone MD CR centers were funded. FSHD was zero percent of NICHD dystrophy funding.

Total funding for FSHD by NICHD decreased by 100% to zero dollars.

Total funding for dystrophy by NICHD increased over the previous year by 1%.

This is due to the shift to a more accurate and better representation of FSHD research from NICHD. It should be an extremely serious concern to children with FSHD.

NHLBI

In fy2006, NHLBI spent $0 on FSHD and $2.3 million on dystrophy. FSHD was zero percent of NHLBI fy2006 dystrophy funding.

Total funding for FSHD by NHLBI remained at zero dollars.

This should be of grave concern as respiratory insufficiency and failure is a growing cause of death in FSHD.

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	$39.9	$1.7	4%
2007	$28,587	$39.9est	$1.7est	4%
2008	$28,587est	$39.6est	$1.7est	4%