Testimony of Daniel Paul Perez, President & CEO, FSH Society, Inc.
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Research on FSHD (Facioscapulohumeral Muscular Dystrophy)
May 1, 2009

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

My name is Daniel Paul Perez, of Bedford, Massachusetts, and I am testifying today as President & CEO of the FSH Society, Inc. (facioscapulohumeral muscular dystrophy) and as an individual who has this common and most prevalent form of muscular dystrophy.

The Need for NIH Funding for FSHD

My testimony is about the profound and devastating effects of a disease known as facioscapulohumeral muscular dystrophy which is also known as facioscapulohumeral muscular disease, FSH muscular dystrophy or FSHD, and the urgent need for increased NIH funding for research on this disorder. According to our research, only a limited amount of work is going on across all the institutes at the NIH. In fact, only 3 of the 27 institutes at the NIH are funding FSHD research, e.g., the National Institute of Neurological Disorders and Stroke (NINDS), the National Institute of Arthritis, Musculoskeletal and Skin Disease (NIAMS), and the Eunice Kennedy Shriver National Institute of Child Health and Human Development (NICHD). Currently, the level of funding from NINDS, NICHD and NIAMS for FSHD research is approximately $3,093,269 (source: NIH RCDC RePORT database system).

Since 1994, I have submitted testimony before both House and Senate Appropriations Committees' subcommittees on Labor, Health and Human Services and Education and Related Agencies which stated that NIH and Congress with modest investments could help bring about a significant research and scientific opportunity which would benefit hundreds of thousands of people worldwide.

Almost, two decades later, I still re-appear before Congress older, wiser and physically much weaker. Again and again, I am asking the Congress of the United States of America to consider the value and merit in supporting FSHD research and why this area of research has been so recalcitrant to modest increases in funding by the NIH. As President of the FSH Society and as the chief activist for the hundreds of thousands of individuals living with FSHD in the United States and worldwide, I will continue to testify year after year arguing the case of wanting to live life free from disease. Hopefully, U.S. Congress and the U.S. NIH will move decisively and proactively to fund FSHD at significantly higher thresholds before this terrible disease takes my life, the lives of my family members affected, and thousands of friends affected by FSHD.

FSHD is a series of errors. The deletion of a stretch of repetitive repeats in the chromosomal structure of the tip of chromosome 4 is the first error; the gene, RNA transcript, or protein that it causes to be mis-expressed is the second, and this primary error is repeated without end. This primary error is the lowest common denominator of the FSHD community e.g. the lesion in chromosome 4. The people living with and involved with FSHD all their lifetime deal with resolving the copious manifestations of this first error. Our lives are a testament to the fact that from this primary error, more complex errors will arise, and that there is no end to the error leading to complexity, and complexity leading to further progression of this debilitating disease,

constant loss of physical ability and in individuals with a moderate to severe course of disease a certain death.

The race to find the gene is an enormous undertaking which traces backward through this series of errors to the starting point. It is a complex race with high expectations and rewards. It is a race in which intellects and creative minds strive to win despite operating on severely reduced research budgets and economies. It is a race that America could win within the next five to ten years if it does not make the error of oversight, omission or of apathy. America could make significant gains in understanding the mechanism of the FSH neuromuscular disorder by investing in new methods and techniques of research. America should realize that now is the time to stop the cascading series of errors. Now is the time to understand the complexity of the situation and, in turn, respond by funding many new FSHD research programs.

The United States of America should not miss this tremendous opportunity that would allow the current generation of people with FSHD, and the many other disorders caused by the same biological mechanism, and their future generations to live life free from disease. The FSHD community has been caught in this never ending circle of error leading to complexity and complexity leading to constant loss of skeletal muscle and functioning. Time is of the essence here. Lives are in the balance and the race against this disease is ongoing. The FSHD community believes that now is the time to move to action and it demands bold and persistent experimentation.

Today, I am asking Congress to communicate to the Public Health Service and National Institutes of Health the need for research funding on the FSHD disorder at a level of $10,000,000 annually in FY2010.

Living with FSHD

The prognosis of FSHD includes both a loss of muscular strength that limits personal and occupational activities, and a total loss of mobility in perhaps twenty percent of the cases. Hearing loss and retinal abnormalities are associated with FSHD.

In fact, I was born to a family that already experienced the extraordinary difficulty of receiving a proper diagnosis for FSHD. In the first few years of my life, I had been diagnosed as having FSHD and a severe hearing loss, which I have come to find out, is part of FSHD.

At 47 years of age, I consider myself a life long survivor of the severe trauma and tension of FSHD, and I do not say this lightly. I have dealt with the continuing, unrelenting and unending loss caused by FSHD from the first second, into the first minute, hour, day, week, over the months, and through the years. Not for a moment is there a reprieve from continual loss of my physical ability; not for a moment is there a time for me to mourn; not for a moment is there relief from the physical and mental pain that is a result of this disease. There is no known treatment and no known cause for this disease.

FSHD has insidiously and systematically deprived me of my childhood, my adolescence, and 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 breathe, the way you relate, the way you parent, the way you love, the way and where you live, the way you drive, and the way people perceive and treat you. I cannot smile; I can no longer hold a baby in my arms; I cannot close my eyes to sleep; I can no longer run or walk on the beach or climb stairs. Every day I am aware of the things that I may not be able to do tomorrow. This is the reality for the twenty-five thousand people living with FSHD in the United States.

The men, women and children who live with the daily consequences of this devastating disease are your friends, neighbors, fellow taxpayers and contributors to the American way of life. With an historical 88% employment rate and an average educational achievement level of 14 years (source: Impairment and Disability Profiles on Neuromuscular Diseases: FacioScapuloHumeral Muscular Dystrophy, Research and Training Center on Neuromuscular Disease, Department of Physical Medicine & Rehabilitation, University of California, Davis and The National Institute on Disability & Rehabilitation Research, 1994), we personally bear our burden of the health care costs and training expenses to prepare for and maintain financial and personal independence.

With quiet dignity, we live our lives as productively as possible with FSHD.

I am here to remind you that FSHD 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.

The Most Prevalent Form of Muscular Dystrophy, Markedly Under-funded at NIH

It is a fact that FSHD is now published in the scientific literature as the most prevalent muscular dystrophy in the world. The incidence of the disease is conservatively estimated to be 1 in 14,285. The prevalence of the disease, those living with the disease ranges to two or three times as many as that number based on our increasing experiences with the disease and more available and accurate genetic diagnostic tests.

The French government research agency, INSERM (Insitut National de la Santé et de la Recherche Medicale) is comparable to the NIH, and it recently published prevalence data for hundreds of diseases in Europe. Notable is the "Orphanet Series" reports covering topics relevant to all rare diseases. The "Prevalence or reported number of published cases listed in alphabetical order of disease" November 2008 - Issue 10 report can be found at internet web site (http://www.orpha.net/orphacom/cahiers/docs/GB/Prevalence_of_rare_diseases_by_alphabetical_list.pdf). This update contains new epidemiological data and modifications to existing data for which new information has been made available. This new information ranks facioscapulohumeral muscular dystrophy (FSHD) as the most prevalent muscular dystrophy followed by Duchenne (DMD) and Becker Muscular dystrophy (BMD) and then in turn myotonic dystrophy (DM). FSHD is historically presented as the third most prevalent muscular dystrophy in the Muscular Dystrophy Community Assistance, Research and Education Amendments of 2001 and 2008 (the MD-CARE Act). This new data ranks FSHD as the first and most prevalent.

Estimated Prevalence (Cases / 100,000)	
Facioscapulohumeral muscular dystrophy (FSHD)	7 / 100,000
Duchenne (DMD) and Becker Muscular dystrophy (BMD) types	5 / 100,000
Steinert myotonic dystrophy (DM)	4.5 / 100,000

For men, women, and children the major consequence of inheriting the most prevalent form of muscular dystrophy, FSHD, is a lifelong progressive and severe loss of all skeletal muscles. FSHD is a terrible, crippling and life shortening disease. No one is immune, it is genetically and spontaneously (by mutation) transmitted to children and it affects entire family constellations.

NIH Muscular Dystrophy Funding Has Tripled Since the Inception of the MD CARE Act

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.

Between fiscal year 2007 and 2008, NIH overall funding for muscular dystrophy decreased as shown in the "Estimates of Funding for Various Research, Condition, and Disease Categories (RCDC)" report on the new Research Portfolio Online Reporting Tool (RePORT) from $58 million to $56 million, a 3 percent decrease. These figures are from the new "2007/2008 NIH Revised Method" columns. The same RCDC RePORT system report shows $47 million as the 2007 figure under the "2007 NIH Historical Method" column, a 23 percent increase and restatement when converting to the new system.

Figures from the RCDC RePORT and the NIH Appropriations History for Muscular Dystrophy report historically provided by **NIH/OD Budget Office & NIH OCPL** show that from the inception of the MD CARE Act 2001, funding has nearly tripled from $21 million to $56 million for muscular dystrophy.

NIH FSHD Funding has Remained Level Since the Inception of the MD CARE Act

Between fiscal year 2006 and 2007, NIH funding for 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).

Between fiscal year 2007 and 2008, NIH funding for FSHD decreased from $4,108,555 to $3 million under the "2007 and 2008 NIH Revised Method." The "2007 NIH Historical Method" was restated to $3 million. In fiscal 2008 under "NIH Revised Method," FSHD was 5.3% of the total muscular dystrophy funding ($3M / $56M). The previous years 2006/2007 figures are revised and restated under "2007 NIH Historical Method" as ($3M / $58M) which is 5.1% of the total muscular dystrophy funding. FSHD funding has merely kept its ratio in the NIH funding portfolio and has not grown in the last seven years.

We highly commend the Director of the NIH on the ease of use and the accuracy of the Research Portfolio Online Reporting Tool (RePORT) report "Estimates of Funding for Various Research, Condition, and Disease Categories (RCDC)" with respect to reporting projects on facioscapulohumeral muscular dystrophy.

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

Fiscal Year	FSHD Research Dollars	FSHD % of MD
2002	$1.3	5%
2003	$1.5	4%
2004	$2.2	6%
2005	$2.0	5%
2006	$1.7	4%
2007	$3	5%
2008	$3	5%

The MD CARE Act 2008 mandates the NIH Director to intensify efforts and research in the muscular dystrophies, including FSHD, across the entire NIH. It should be very concerning that in the last seven years muscular dystrophy has tripled to $56 million and that FSHD has remained at

five (5) percent of the NIH muscular dystrophy portfolio or $3 million. Only three of the institutes at the NIH are funding FSHD. OD, NHLBI, NIGMS, NIBIB, NIDCD, NHGRI, NEI, NIA, 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.

Now, FSHD is published as the most prevalent muscular dystrophy, and given the extraordinary interest of the scientific and clinical communities in its unique disease mechanism, it defies gravity that it still remains the most prevalent and one of the most underfunded dystrophies at the NIH and in the federal research agency system (CDC, DoD and FDA). In 2008, the third most prevalent dystrophy, Duchenne (DMD) and Becker Muscular dystrophy (BMD) type, received $22 million from NIH. In 2008, the second most prevalent dystrophy myotonic dystrophy (DM), received $9 million from NIH. In 2008, the most prevalent dystrophy, facioscapulohumeral muscular dystrophy (FSHD), received $3 million from NIH. It is now time to flip the stack and to make sure that FSHD with its equal burden of disease and highest prevalence gets more funding, stimulus and that NIH program staff initiates request for applications specifically in FSHD. It is crystal clear, if not completely black and white, that the open mechanism program announcement and investigator driven model are not achieving the goal mandated by the MD CARE Acts 2001/2008 and by the NIH Action Plan for the Muscular Dystrophies as submitted to the Congress by the NIH. Efforts of excellent program staff and leadership at NIH, excellent reviewers and study sections, excellent and outstanding researchers working on FSHD and submitting applications to the NIH, and extraordinary efforts of the volunteer health agencies working in this area have not yet enabled FSHD funding to increase at the NIH. It is time for requests, contracts ands calls for researcher proposals on FSHD to bootstrap existing FSHD research worldwide.

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.

Our request to the NIH Appropriations Subcommittee

We request this year in FY2010, immediate help for those of us coping with and dying from FSHD. We ask NIH to fund research on FSHD at a level of $10 million in FY2010.

We implore the Appropriations Committee to request that the Director of NIH, the Chairman/Chairwoman, and Executive Secretary of the federal advisory committee Muscular Dystrophy Coordinating Committee mandated by the MD CARE Act 2008, to increase the amount of FSHD research and projects in its portfolios using all available passive and pro-active mechanisms and interagency committees. Given the knowledge base and current opportunity for breakthroughs in treating FSHD it is inequitable that only three of the twelve NIH institutes covering muscular dystrophy have a handful of research grants for FSHD. 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. Our successes are continuing and your support must continue and increase.

We ask you to fund NIH research on facioscapulohumeral muscular dystrophy (FSHD) at a level of $10 million in FY2010.

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