

Name of the Organization Submitting Testimony:

**FSH Society, Inc.**

Subcommittee for which the Testimony has been Prepared:

**Labor, Health and Human Services, Education and Related Agencies**

Department and/or Agency the Testimony is Addressing:

**National Institutes of Health (NIH)**

**Testimony of Daniel Paul Perez, President & CEO, FSH Society, Inc.  
telephone: (781) 275-7781, e-mail: daniel.perez@fshsociety.org before the  
U.S. Senate Appropriations Subcommittee on  
Labor, Health and Human Services, Education and Related Agencies on the  
Subject of FY2010 Appropriations for National Institutes of Health (NIH)  
Research on Facioscapulohumeral Muscular Dystrophy May 25, 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 and 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 National Institutes of Health (NIH). In fact, only three (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 U.S. House and U.S. 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.

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**

As a man with facioscapulohumeral muscular dystrophy, I will tell you that it is a hard way to live, and that FSHD is a strong fort—it will last a lifetime. Unless Congress mandates that the NIH to ensure that it receives sufficient grant applications of highest quality on FSHD and to spend an equitable ratio of NIH muscular dystrophy dollars on FSHD which is now conservatively ten (10) million dollars.

At 47 years of age, I consider myself a lifelong 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.

Look at what this disease does to people. Look at us. Look at what we see—a child with a profound hearing loss, the broken innocence of a child, alienation at an early age, a decision not to marry, a decision not to have biological children, disability in the prime of life, incapacitation in middle age, the guilt of a parent, a lifetime of physical challenge, a suicide, a premature death, anxiety caused by uncontrollable loss, decades spent somewhere between the able and the disabled, the loss of ambulating, the unstoppable atrophy and loss of muscle and the humiliation endured in the process.

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.

### **The Most Prevalent Form of Muscular Dystrophy is now 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](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

### **NIH Muscular Dystrophy Funding Has Tripled Since the Inception of the MD CARE Act (\$21M to \$56M)**

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 (\$3M / \$56M)**

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**

<b>Fiscal Year</b>	<b>FSHD Research Dollars</b>	<b>FSHD % of MD</b>
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 NIH requests, contracts and 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 and a reauthorization 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.