We're happy to announce the 10th anniversary of the National Registry! This anniversary represents 10 years of connecting patients and researchers. Your dedication is a great example of the teamwork needed to advance research in myotonic dystrophy (DM) and facioscapulohumeral muscular dystrophy (FSHD). Such teamwork involves discussions amongst physicians, scientists, research support staff, patients, and family members. In a way, we are all members of a "research family," each of us learning from each other, hitting bumps and success along the way, and more importantly, growing together and reaching for a common goal. After 10 years of growth, the National Registry is one of the largest resources available to investigators in the world to study DM and FSHD.

We appreciate all that our members have done to make the Registry a success – such as completing all the paperwork to join the Registry, referring family members to join, completing annual updates year after year, and participating in the research studies that we announce. Helping the Registry in any or all of these ways allows researchers to gain a greater understanding of DM and FSHD.

Through this newsletter, we'd like to share some of the Registry's recent accomplishments that you helped make a reality. This includes new observations about the symptoms and progression of DM and FSHD based on information reported by Registry members. We will also describe recent studies that Registry members have participated in as well as new research opportunities that will be offered to Registry members. Lastly, we'll highlight information on issues including sleep and pain management, and we'll share our plans for the coming years.

In the coming years, we anticipate exciting developments in the research and clinical care of DM and FSHD. Researchers have made significant advances in explaining the causes of symptoms of DM and FSHD, and understanding the cause of a symptom is a critical step towards treating it. Experimental treatments for DM and FSHD are on the horizon, and, when available, the Registry will be a vital tool for researchers to use for contacting and recruiting patients for clinical trials. It is also an important source

Registry partners with University of Rochester Wellstone MDCRC

The National Institutes of Health (NIH) established Centers of Excellence in muscular dystrophy research in 2003, in honor of the late Senator Paul D. Wellstone of Minnesota. Senator Wellstone was a strong supporter of medical research and helped support many laws in Congress to expand research opportunities into muscular dystrophies.

The NIH currently funds six Senator Wellstone Muscular Dystrophy Cooperative Research Centers (MDCRC). The goals of the MDCRC are to study, diagnosis, and develop new treatments for people with muscular dystrophy. The MDCRC are designed to share medical technologies and expertise, and the Centers teach and train new researchers in the muscular dystrophy field. The Centers funded to date are listed to the right.

Current and past Wellstone MDCRC

Year: 2008 to 2013
- Boston Biomedical Research Institute
- University of North Carolina
- University of Rochester

Year: 2005 to 2010
- Children's National Medical Center
- University of Iowa
- University of Pennsylvania and Johns Hopkins University

Year: 2003 to 2008
- University of Pittsburgh
- University of Rochester
- University of Washington, Seattle

University of Rochester Wellstone MDCRC

Years 2003-2008

The University of Rochester Wellstone MDCRC was designed to integrate basic, translational, and clinical research on the two most common forms of adult muscular dystrophy, DM and FSHD.

Project 1 (RNA Toxicity) used mouse and other cell models to explore the biology of myotonic dystrophy type 1 (DM1).

Project 2 (Dose Escalation Trail of IPLEX in DM1) demonstrated preliminary evidence that daily subcutaneous injections of IPLEX [insulin-like growth factor-1 (IGF1) complexed with recombinant IGF binding protein-3] were safe and well tolerated in DM1.

Project 3 (Biological mechanisms of FSHD) provided detailed analyses of muscle tissue samples in patients and has increased our knowledge of the molecular and cellular pathophysiology of FSHD.

Years 2008-2010

Scientific Core: "Repository and National Registry"

The purpose of the Repository is to distribute research reagents and biological materials to investigators. The National Registry provides a mechanism to establish contact between people with DM and FSHD and researchers.

Project 1: "Pathogenesis and progression in Myotonic Dystrophy"

The Aims of this project are to evaluate bench to bedside aspects of disease progression. The goals of project are to study the genetic instability of DM1, to study the relationship between various biochemical markers of DM1 and muscle impairment, and to conduct a longitudinal study of a large cohort of DM1 patients to determine which outcome measures are most sensitive to monitor disease progression and suitable as endpoints in clinical trials.

Project 2: "Experimental Therapy of Myotonic Dystrophy"

The Aims of this project encompass gene therapy, oligonucleotide therapy, and small molecule (drug) therapy for myotonic dystrophy.

The resources of the University of Rochester Wellstone MDCRC have led to significant progress into the understanding of potential pathomechanisms of FSHD. These resources have also created a surge in DM research and, we believe, a very favorable hope for effective treatments.

Both myotonic dystrophy type 1 (DM1) and type 2 (DM2) share many similar symptoms. Examples of shared symptoms include trouble relaxing muscles (myotonia), cataracts, and heart abnormalities. There are also many differences in symptoms between the two DM subtypes. DM1 is characterized by more weakness in the distal parts of the body (hands and feet) compared to more proximal weakness (hips and upper legs) in DM2. DM1 can also result in a severe presentation at birth (congenital form) that is not seen in DM2.

Early-onset DM1

A small subset of individuals with DM1 exhibit symptoms before adulthood. Those with congenital DM1 have severe symptoms of the disease at birth, and others with childhood-onset DM1 show symptoms of the disease during childhood, or before about 11 years of age. Little information is available on childhood-onset DM1 and whether or not symptoms, progression of the disease, and burden of disease are unique for these patients.

Data from the Registry showed that 23 childhood-onset members and 33 congenital members had enrolled as of September 2009. Childhood and congenital members represented 3.5% and 5.1% of all DM1 members enrolled in the Registry.

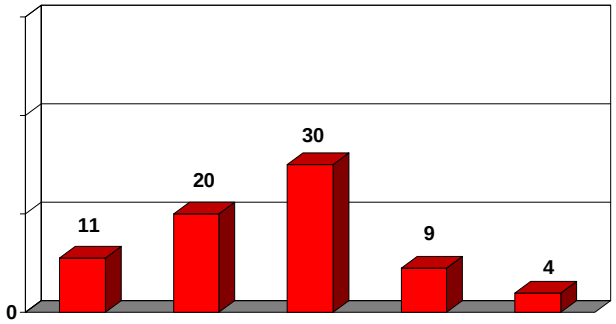
Members with childhood-onset DM1 reported a greater variety of first symptoms, possibly indicating greater variability between these patients in how their first symptoms develop. Registry members with childhood-onset DM reported less use of physical, occupational, and speech therapies, but greater use of psychological counseling and more frequent occurrence of psychological disorders. Attention deficit/hyperactivity disorder (ADHD) was the most common psychological disorder for both congenital and childhood-onset members. Further research is needed to

Presentation: Standards of care and the management of FSHD patients

Registry staff presentation at the 171st European Neuromuscular Centre (ENMC) International Workshop, titled, "Standards of care and the management of FSHD patients".

The goal of this ENMC conference was to develop "standards of care" and outline goals to develop new treatments for FSHD. Use of the Registry helped to reach these goals!

We presented anonymous information from the Registry about patient demographics, diagnostic test results, and how the Registry operates. Such information may help scientists around the world better understand how to develop registries in their countries and how various registries can compare and share information.



We also presented information on the use of rehabilitation therapies and assistive devices of FSHD members. The graph indicates the percentages of FSHD subjects who reported using assistive devices at enrollment. Canes were the most commonly used assistive device and were used by 30% of FSHD members.

About 30 scientists from around the world attended this conference to discuss these results and other information from various research studies and physician groups. The leaders of this conference are summarizing the meeting and plan to publish clinical guidelines for FSHD that may help doctors better treat patients worldwide! The leaders of this conference will also develop a list of key questions in need of further research. A publication and more information is pending soon!

This conference was co-sponsored and led by collaborators with the ENMC and the Fields Center for FSHD & Neuromuscular Research.

Publication: Factors that may influence chronic pain in DM1 and FSHD patients

Investigators recruited members of the National Registry to participate in their study to measure how DM1 and FSHD patients may respond to pain in terms of biology, psychology, and social factors. Investigators were from the University of California at Davis, University of Washington, and Rovira I Virgili University of Spain.

A total of 395 questionnaires were mailed to interested patients. About 75% of these research subjects were members of the Registry (296 of 395 subjects). Individuals who reported experiencing pain at the time of the study or in the previous 3 months were included in the study (a total of 182 DM1 and FSHD patients). Study participants completed a survey about a variety of questions related to pain.

Investigators found that feelings of guilt and self-blame increased chronic pain in DM1 and FSHD. Pain significantly interfered with daily activities and decreased psychological health in patients with poor social support (from family members, friends, etc). Pain was also worsened in individuals who thought about their symptoms repeatedly and had feelings of helplessness.

Results indicated that patients with more social support or access to therapies had decreased burdens of pain in daily living and better psychological well being. Additional studies are needed to develop better treatments and therapies for chronic pain in DM1 and FSHD and to measure pain in DM2.

"Patients with higher social support or perceived access to therapies were associated with decreased burdens of pain in daily living and better psychological well being.."

Reference Title: Impact of biopsychosocial factors on chronic pain in persons with DM and FSHD.

Research journal: Am J Hosp Palliat Care. 2009 Aug-Sep; 26(4):308-19

This research was supported by the National Institutes of Health, National Institute of Child Health and Human Development, National Center for Rehabilitation Research (grant no. P01HD33988), and the National Institute for Disability Rehabilitation Research (grant no. H133B031118).

Investigators recruiting members of the Registry for studies

Survey of symptoms most important to DM and FSHD patients

Study investigator: Dr. Chad Heatwole, University of Rochester (Rochester, NY); study supported by the Muscular Dystrophy Association

Investigators are surveying patients about issues and symptoms that were previously identified by DM and FSHD patients as being important to their daily lives. The researchers are recruiting a large number of patients to provide further insight into these areas. The National Registry will send recruitment letters to all eligible members of the Registry over the next several months. Members will then have the opportunity to complete the survey. The survey will take approximately 15 minutes (or less) to complete and all responses will be strictly confidential.

Update from the study team: In April 2010, 530 surveys were sent to eligible DM1 registry participants. We very much appreciate all of the responses we have received so far. Thank you for your partnership with this research! We will continue to accept survey results over the upcoming weeks to give an opportunity to those participants who have not yet completed the survey to contribute to this research. Surveys for FSHD and DM2 are being developed and pending approval by our University's Research Subjects Review Board and will be mailed in the near future. Your input will greatly assist us in the understanding of this disease and help us with future re

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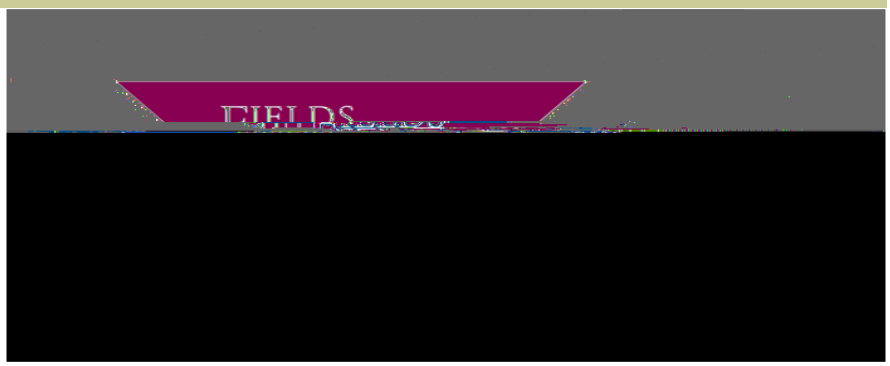
Fields Center for FSHD & Neuromuscular Research



Dear Members of the National Registry,

THANK YOU for your enthusiastic support for the work of the Fields Center for FSHD & Neuromuscular Research! A year ago, we asked you to consider volunteering for a study being conducted at the University of Rochester. BOY, did you volunteer! Over the past year, we've talked with hundreds of Registry members about what the Fields Center is doing. This may be a good time to update the entire membership about what is happening as well.

The Fields Center will celebrate its third birthday this summer. That time has flown by! Since we began, we reached out to you to offer two different types of opportunities:

> Scientific opportunities, with requests that you work with Fields Center researchers by participating in the "CAMP Study" (the Cellular and Molecular Pathophysiology of FSH Dystrophy) and the "PEVA Study" (the Position Effect and Vascular Adaptation of FSH Dystrophy), and
> Educational opportunities, with invitations to our Annual Fields Center Patient Days.

Here is an update on our partnership:

> Where we are: We were pleasantly overwhelmed by your interest in the CAMP study! Within the first month of our announcement, over 125 Registry members called to volunteer from places as close as Rochester, NY and as far away as Alaska and Hawaii! AND, you kept calling! AND, you KEEP calling! Thank you! We are grateful for the generosity of those people who shared their information and blood and tissue samples, whether you came to the UR to participate or whether you helped us from your home community. It has been a delight working with all of you and we are sincerely grateful for your support.
>
> Going forward: We are continuing to recruit people for this study. A special message for people that have already volunteered: ***We still need you!*** We are still processing records and scheduling visits as quickly as we can. Thank you for your patience with us — we will be getting back in touch with you. If you have questions about your status or to hear more about this study, please call us at **585.275.7680**.
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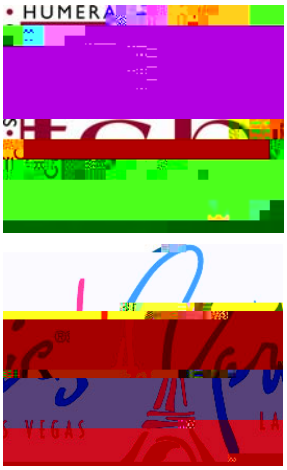
> The Fields Center Directors - Dr. Rabi Tawil and Dr. Silvere van der Maarel — will be hosting our 3rd Annual Fields Center Patient Day in Rochester, NY on Saturday, September 18, 2010. Our 2008 and 2009 teaching days were fun and informative get-togethers that let our patients, scientists and clinicians share information in a casual setting. Again this year, we will gather at the Memorial Art Gallery. This year promises to be an especially exciting event — please join us! There is no cost for the meeting, parking, or meals and admission to the Art Gallery is free to attendees. Just call us to register. We'd love to see you there!

The Fields Center investigators and staff are grateful for your interest and active participation. It is only with the active involvement of patients and families that we move forward toward our shared goal – to develop effective treatments for people with FSHD. THANK YOU for being part of the progress!

For more information about the Fields Center, please contact us in the way most convenient for you: by phone: **585.275.7680**, or by e-mail: FieldsCenter@urmc.rochester.edu. You can also visit our website at www.FieldsCenter.org to find information about our studies and links to online publications and resources for people with FSH Dystrophy. Please keep in touch!

Study investigator:s Dr. Lisa Kalman, Center for Disease Control and Prevention (Atlanta, GA); Dr. Richard Moxley, University of Rochester (Rochester, NY) & Dr. Lorraine



The FSH Society, Inc is a network of patients, family members, and researchers dedicated towards advancing understanding of FSHD and developing new treatments. One example is how the FSH Society funded an important study that recruited many member of the Registry. This study measured pregnancy and birth outcomes in FSHD and was published in the medical journal, Neurology (2006 Nov 28; 67(10):1887-9).

Colleagues of the FSH Society, Inc will be discussing many research opportunities and important topics related to FSHD at their upcoming Patient/Researcher Meeting. The FSH Society's 2010 International Patient/Researcher Network Meeting will be held at the Paris and Bally's Hotels in Las Vegas, July 30-August 1. The conference will begin with registration and lunch at noon on Friday, July 30, and conclude after lunch on Sunday, August 1. The full program, meeting registration, and hotel reservation information are available on the Society website, www.fshsociety.org.

Small group discussions will enable informal sharing by patients and family members, around such topics as:

* Caregivers: Sharing triumphs and trials
* Leisure Time and the Freedom to Travel
* Taking Stock of Your Future in the Workplace
* Maintaining a Good Diet and Good Nutrition
* The Expert Patient: Managing Dialog with Your Physician
* Advocacy and Disability Rights

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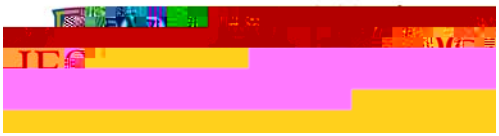

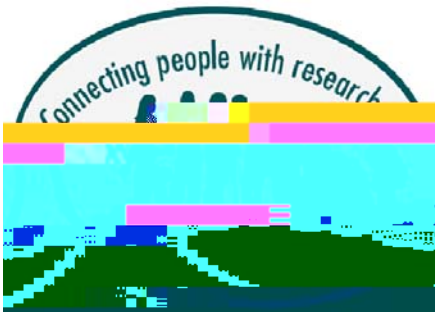
Connecting people with research