This newsletter comes to share the exciting plans and achievements of the National Registry and to emphasize progress being made in research and clinical care of myotonic dystrophy (DM) and facioscapulohumeral muscular dystrophy (FSHD). Some of this progress is a direct result of your participation in our National Registry and comes from the information on your patient information forms and your annual updates.

Based upon the results of clinical information that you have provided to the Registry and upon data from other studies, we are gaining a better knowledge of how the different symptoms of your disease progress and what may be the most important features to measure in determining the effectiveness of new treatments. Advances in research on the gene defects responsible for DM (DM1 & DM2) and FSHD have led to better understanding of the causes of specific disease symptoms in both DM and FSHD and have given increased optimism about potential new therapeutic approaches. We and other investigators are also carrying out studies to better understand how your symptoms change over time, what symptoms matter most to you and to your family members, and how to better measure your symptoms in research studies and in your doctors' offices. This information is not only important to you and your care providers but it is also important to the companies that will help develop and establish new treatments. Companies want to know symptoms that matter most to patients, estab-

Registry updates

We have now enrolled over 1450 members into the National Registry! *But what does the Registry represent?* The Registry represents patients from all 50 states, and what we believe is the largest and most diverse population of individuals with myotonic dystrophy (DM) and facioscapulohumeral muscular dystrophy (FSHD).

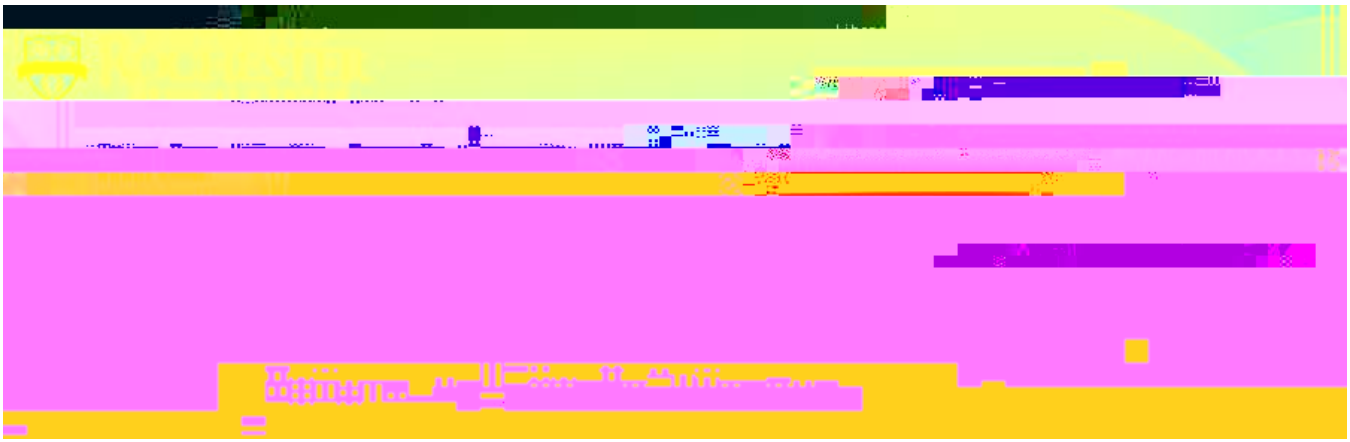
But more importantly, the Registry represents over 1,450 motivated patients and family members who are dedicated to research and gaining a greater understanding of DM and FSHD. It contains information about your muscle weakness, symptoms, therapies, medications, and assistive devices. For members who enrolled when the Registry began and completed each annual update, the data represents over 7 years of tracking your symptoms. For all of you, we hope the Registry gives voice to your concerns and hopes.

The collective information in the Registry continues to help guide researchers and clinical care providers to a better understanding of your disease manifestations and helps to stimulate and develop research studies. There have been 5 Registry based papers published in medical and scientific journals to date. These papers contain information about the Registry and its members. We and other investigators have also presented 13 posters and lectures on information contained in the Registry. Investigators have reported information to describe: the pain that commonly occurs in DM and FSHD patients, the problems with excessive sleepiness that trouble some patients with DM, the outcomes of pregnancy in mothers with FSHD, and the molecular biology of each disease.

We hope you share our excitement about these accomplishments you have helped researchers achieve and that you are looking forward to our future goals as we continue to grow the Registry with new members and assist researchers as they pursue new studies.

The National Registry currently has 1456 members (see Figure 1).

Every year, we send out questionnaires (annual updates) to all Registry members. The information you provide in your annual updates extends and amplifies our understanding of your disease. These annual updates document how your symptoms progress, how your medications change, and how other medical problems may develop or improve. This information is essential as we develop new treatments since some therapies may slow down, control or cure only one or two disease manifestations. Researchers, clinicians, and the pharmaceutical companies need this annual update information to know how to design and evaluate trials of treatment. These annual forms also help us keep track of your contact information if you move, change phone numbers, or change jobs. Even if nothing has changed over the past year, it is still helpful to document that your symptoms or other conditions have remained



Don’t have internet access?

If you don’t have internet access, but are interested in the information provided on our website, please contact our team and we would be happy to mail you this information.

Research Updates

Many members of the National Registry often tell us that they would love to participate in more studies. We are encouraged by your enthusiasm and your continued interest in joining research studies! We also realize the frustration of members who would like to participate in clinical or drug trials, but who can not enroll into these studies for various reasons. Many factors hinder the development of large clinical studies that could include everyone who has an interest in participating. One major factor is our limited understanding of the exact cause for the different symptoms that trouble patients with DM and FSHD at various stages of these diseases. This limitation has stifled identification of promising therapeutic approaches. Currently only a few studies develop each year for DM or FSHD and the inclusion criteria and the cap on the number of patients often make it impossible for everyone who is interested to participate. Other restrictions include reduced availability of grant money increased costs of travel, and increased costs of study supplies and tests. In addition, it is often a lengthy and challenging process to "translate" or understand how the biology or drugs tested in the laboratory affect humans. We and others in the muscular dystrophy field are working to overcome the challenges associated with developing clinical trials and are optimistic that in the near future there will be a significant increase in opportunities for members of the Registry to participate in trials of treatment.

"One important feature of the National Registry is that it allows <u>all</u> members to participate in research."

One important feature of the National Registry is that it allows **all** members to participate in research. Patients can participate regardless of the specifics of their disease and their ability to travel. **All members of the Registry help us collect vital information about your disease manifestations through questionnaires and postal mailings.**

Types of Research Questions

While questionnaires from our team and other investigators may not document all of your symptoms, they are the first steps to better understand DM and FSHD and to develop future, larger studies. Our questionnaires provide an overview and specific information about your diseases that may guide the development of new research studies. Additionally, the answers you give to the annual updates and to questionnaires from researchers studying members of the Registry have important implications for the clinical care decisions made by medical doctors. For example, do DM1 and DM2 patients have similar manifestations of these two forms of the disease? Which symptoms are most important to DM and FSHD patients and family members? What are the exact biological reasons of why FSHD and DM occur? How can we use this information to better manage and treat your disease?

These questions are only a small sample of many exciting research questions that provide opportunities for investigations being pursued by researchers around the world. As more information is gained about DM and FSHD, we are confident that additional and larger studies will develop.

Current accomplishments and future goals

We are eager to report many of the current accomplishments and future goals of the National Registry in this newsletter. Highlights described in more detail on the following pages include:

Spanish translation: Plans are described to translate our forms and sections of our website into Spanish.

Scientific papers: Published papers that have recruited members of the Registry to participate in clinical studies are summarized.

Data reports: Common symptoms of Registry members are described and how these manifestations progress each year;

Current studies: Clinical research studies in Minnesota and New York that are currently recruiting eligible Registry members are announced;

Future studies: Studies being developed to recruit members of the National Registry in the near future are announced.

Spanish translations

Inclusion of potentially underserved patients ….will make research results more representative, and therefore more accurate.

We and others investigators are concerned that there may be a significant population of Spanish speaking patients in the United States that are underserved in obtaining information about and opportunities to participate in research due to language barriers. This concern is important because such individuals may benefit from participating in research. Inclusion of potentially underserved patients (for example, those patients with language barriers, severe symptoms, those unable to travel) will make research results more representative, and therefore more accurate. As mentioned previously, Registry members are quite diverse, however taking steps to further diversify the Registry will help us better understand your disease. To begin to ease language barriers in the Registry, we are continuing to develop plans to translate our forms and sections of our website into Spanish.

We hope to complete the translation of the Registry forms this year. Once our forms are approved, we will mail recruitment letters to neurologists and other neuromuscular specialists across the country to promote enrollment of patients whose first language is Spanish.

Opportunities may exist in the future to develop the National Registry forms into other languages. Information about the translation of our forms to Spanish and potentially other languages will be updated on our Registry website and provided in our next newsletter.

Published papers that reference the Registry

The information collected by the National Registry is continually used by researchers and the Registry staff to explore a variety of topics and add to the growing body of knowledge on myotonic dystrophy and facioscapulohumeral muscular dystrophy. The results of their work are presented at research conferences, government sponsored health and research workshops, meetings for patient support groups, and in scientific journals.

Recently published papers that reference the Registry are described on the following page.

Background: Gastrointestinal (GI) problems are a frequent and serious complaint in DM1 patients. The cause of GI disturbances in DM1 remains unclear and limited information is available about the

More detailed descriptions and cont

Future studies recruiting Registry members

- **Effects of Aging on Patients with Muscular Dystrophy and Other Disabilities**

The National Institute on Disability and Rehabilitation Research has recently funded a Research and Training Center (RRTC) at the University of Washington's Department of Rehabilitation Medicine. The purpose of this Center is to study the challenges faced by those aging with muscular dystrophy and other physical disabilities.

Research activities conducted by the RRTC provide an exciting way to contribute to knowledge about aging with muscular dystrophy. The utilization of a questionnaire to better understand the scope of aging with muscular dystrophy is just one example of this Center's upcoming projects.

- **Pathogenesis and Progression in Myotonic Dystrophy**

The National Institutes of Health has refunded the University of Rochester Medical Center as one of its six Senator Paul D. Wellstone Muscular Dystrophy Cooperative Research Centers. One of the many purposes of the University of Rochester Wellstone Center is to study the causes of muscle weakness and stiffness (myotonia) in myotonic dystrophy.

This study, led by Dr. Richard Moxley, will also attempt to better understand how myotonic dystrophy changes and progresses over time. A thorough understanding of the natural progression of myotonic dystrophy is necessary for researchers to plan and develop clinical studies and treatment trials.

This study will enroll individuals with myotonic dystrophy type 1, myotonic dystrophy type 2, and unaffected volunteers. Patients with myotonic dystrophy types 1 and 2 will be asked to participate in three visits to the University of Rochester. During these visits, patients will take part in a series of evaluations, including muscle strength testing, DEXA scan to measure muscle mass, chest x-ray, electrocardiogram (EKG), blood sample collection, and a thin needle muscle biopsy.

More detailed descriptions and contact information to participate in these studies will be sent to eligible Registry members in the near future.

Resources

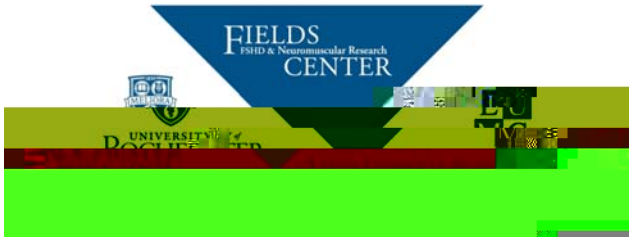
The following section provides educational information and announcements of events related to muscular dystrophy. Sources of information for this part of the newsletter include the National Institutes of Health, the Fields Center at the University of Rochester, and the Research and Training Center (RRTC) at the University of Washington.

All organizations featured in our newsletters and on our website have either sponsored the Registry or have conducted or planned research utilizing the resources of the Registry. Information from these resources does not represent the Registry, but, in general, fits with the goals of the Registry. We welcome past, present, and future collaborators to use the resources of the Registry to disseminate pertinent, educational information to Registry members.

NIH Links

Per the NIH website: "The National Institutes of Health (NIH) is the primary Federal agency for conducting and supporting medical research. Helping to lead the way toward important medical discoveries that improve people's health and save lives, NIH scientists investigate ways to prevent disease as well as the causes, treatments, and even cures for common and rare diseases."

- National Institute of Arthritis and Musculoskeletal and Skin Diseases (NIAMS): http://www.niams.nih.gov/
- National Institute of Neurological Disorders and Stroke (NINDS): http://www.ninds.nih.gov/
- NIH Senator Paul D. Wellstone Muscular Dystrophy Cooperative Research Centers: http://www.wellstonemdcenters.nih.gov/index.htm



Save the Date!

Please join us for our 2nd Annual Patient Day
Friday, September 25, 2009

The Partners of the
Fields Center for FSHD & Neuromuscular Research
cordially invite you to the

2nd Annual Fields Center Patient Day!

Friday, September 25, 2009
9:00 AM – 3:30 PM

Featuring internationally known researchers
discussing topics requested by patients and family members.
Admission and parking are free. Lunch will be provided.

For additional information or to pre-register, please call
Karen Richards at 585.275.6372
or
contact us by email at
FieldsCenter@urmc.rochester.edu

We look forward to seeing you there!

Fields Center for FSHD & Neuromuscular Research
University of Rochester Medical Center
Department of Neurology
601 Elmwood Avenue, Box 673
Rochester, NY 14642
Phone #: 585.275.7680 or 585.275.6372
Email:FieldsCenter@urmc.rochester.edu
Web: www.FieldsCenter.org

Aging well with muscular dystrophy

Guest column, from colleagues at the University of Washington:

Growing older comes with a number of benefits, including increased wisdom and experience. But growing older also can pose challenges that can limit participation in many valued life activities. These challenges can be particularly difficult in people who have muscular dystrophy (MD), who not only have to face the effects of aging, but who continuously deal with the symptoms associated with MD, such as muscle weakness, pain, and fatigue. Problems associated with aging can include increased sleep and mood disturbance. Researchers have not given adequate attention to identifying, developing, and testing effective treatments for these problems in people with muscular dystrophy.

To address this gap in understanding, the National Institute on Disability and Rehabilitation Research has recently funded a Research and Training Center (RRTC) at the University of Washington's Department of Rehabilitation Medicine.

Purpose of RRTC

The purpose of this Center is to study the challenges faced by those aging with MD and other physical disabilities. The target objectives of the RRTC are to:

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Disclaimer

Contact Us

The National Registry
University of Rochester Medical Center
601 Elmwood Avenue, Box 673
Rochester, NY 14642

Toll Free Phone: 1-888-925-4302
Local Phone (Rochester, NY): 585-276-0004
Fax: 585-273-1255
E-mail: dystrophy_registry@urmc.rochester.edu

dystrophyregistry.org

This project has been funded in whole or in part with Federal Funds from the National Institute of Arthritis and Musculoskeletal and Skin Diseases (NIAMS), National Institute of Neurological Disorders and Stroke (NINDS), National Institutes of Health, Department of Health and Human Services, under Contract # NO1-AR-5-2274.

The Registry team

We have enjoyed getting to know patients and family members enrolled in the Registry over the past 7 years, and we would like to share information about our interests and background with you. The leadership and staff listed below have enjoyed writing this newsletter and appreciate your ongoing dedication to research.

Dr. Richard Moxley, III is an established investigator with a long history of expertise and international reputation in research in neuromuscular disease and in the development and implementation of clinical trials in muscular dystrophy. He has been with the URMC Department of Neurology for over 30 years and has won numerous awards from patient advocacy groups and scientific and medical organizations.

Jim Hilbert is a research coordinator with a background in biology and a master's degree in exercise science. He has assisted the development of the Registry for the past 5 years and has presented data from Registry at national and international conferences about tumors in DM, infantile and childhood FSHD, and the progression of disease symptoms.

Liz Luebbe is a research coordinator with a background in psychology and is currently pursuing a master's degree in counseling. She has been with the Registry team for almost a year and is eager to pursue studies using data from the Registry to analyze psychological issues in DM and FSHD and to study various burdens of disease.

Thank you for your support!

We are happy to answer any of your questions and welcome your opinions, ideas, and suggestions for the Registry. Please call us toll free at (888) 925-4302.