National Registry of DM and FSHD Patients and Family Members

Volume 6 April 2009

Special points of interest:

- Enrolled 1,456 members to date.
- Published reports based upon Registry member information, such as, gastrointestinal symptoms in DM, pain in DM and FSHD, excessive sleepiness and the biology of each disease.
- Approved 2 new studies to recruit members of the Registry.
- Introduced a new column: "Aging well with muscular dystrophy."

Inside this issue:

Introduction

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, establish their reliable measurement, and show that the new therapies control these symptoms.

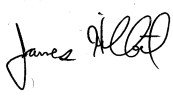
The Registry is poised to assist researchers, different funding agencies, and pharmaceutical companies in carrying out these studies. We need to continue the growth of our Registry membership and continue to increase the information in our database. All members of the Registry are vital to our success and each member plays an important role in helping us better understand all the symptoms of DM and FSHD.

New observations based upon your Registry data and other updates on information about research in DM and FSHD are the primary features of this newsletter. We hope you share our excitement about the valuable data that you have provided to the Registry, and that you are pleased with the growth of the National Registry, especially with its enrollment of over 1,450 members. We also hope that as you read our newsletter, please note that through your contributions to the database in the Registry, you are playing a vital role in advancing knowledge of your disease! We further hope that the newsletter increases your interest in participating in clinical studies now and in the future.

Thank you again for your continued support to help us grow and strengthen the "Registry family."

Best wishes for a healthy and happy year!

Richard T. Moxley, III, M.D.
Principle Investigator



James Hilbert, M.S.
Research Coordinator

Elizabeth Luebbe
Research Coordinator

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.

"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.

Enrollment and annual updates

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 stable. And we just like hearing from you!

Figure 2 indicates a high return rate of the annual updates. The graph shows that the highest response rate occurs after one year of enrollment, when approximately 80% of members returned their forms. We would like to keep these return rates very high, so please keep up the great work!

Figure 1:

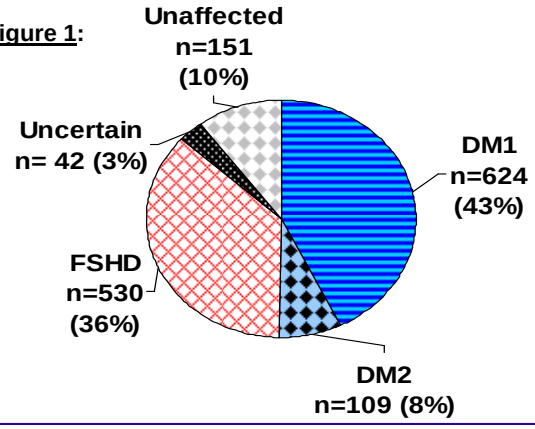
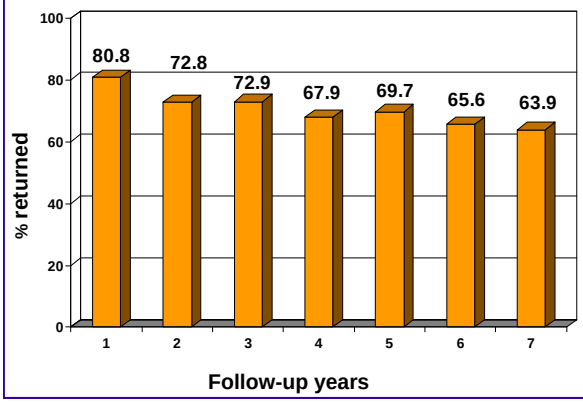


Figure 2: Return rate of annually updated forms.



Website Updates

Our website has a new look! If you've visited our website recently, you may have also noticed that the website has been updated to highlight some of our latest accomplishments and goals. These updates include:

New Research Information: Read about recent advances in DM and FSHD research from studies that involved Registry members. Registry members made a difference in these studies!

Website Links: Explore resources for clinical trials and access extensive medical and health-related information through National Institutes of Health (NIH) websites.

Registry Newsletters: Read, download, or print any Registry newsletter from our website.

Coming soon to our website:

Online Packet Request: Do you have affected or unaffected family members or friends with DM or FSHD that might want to join the Registry? Individuals interested in joining the Registry will soon be able to request an enrollment packet simply by entering their contact information in a brief form online. The information in this form is sent directly to the Registry staff via a **secure internet connection**. Once a request is received an application packet is mailed to the individual at the address provided.
If family members, friends, and others with DM and FSHD are interesting in applying now, they can contact us toll free at 1-888-925-4302 or by emailing us at dystrophy_registry@urmc.rochester.edu

Aging Well with Muscular Dystrophy Informational Columns: Our colleagues from the University of Washington are teaming up with us to provide new, relevant medical information to patients with muscular dystrophy. Our website will be updated every three to four months with new information. These new columns will be developed based on current research and based on the expertise of the team at the University of Washington. *Please see page 11 for more information.*

To see these updates and other information, please visits our website!

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

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

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 **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.

Excessive Daytime Sleepiness (EDS) in DM (2008)

Ciafaloni, E., Mignot, E., Sansone, V., Hilbert, J. E., Lin, L., Lin, X., Liu L. C., Pigeon W. R., Perlis M. L., Thornton C. A. (2008). The hypocretin neurotransmission system in myotonic dystrophy type 1. Neurology, 70 (3), 226-230.

"The authors conclude that excessive sleepiness in DM1 is different from the sleep disturbance that occurs in narcolepsy."

Paper published by Investigators at the University of Rochester and colleagues at Sanford University and the University of Milan.

Excessive daytime sleepiness (EDS) is a common and often significant symptom for patients with myotonic dystrophy type 1 (DM1). Members of the Registry were recruited to participate in a study to measure such symptoms. Other investigators have reported that narcolepsy-like sleep-onset REM has been observed in some patients with DM1. Similarly, the hypocretin (Hcrt) neurotransmission, which is impaired in narcolepsy, was reported previously to be reduced in DM1.

The current study further investigated the Hcrt neurotransmission system in DM1 for patients with and without EDS. Patients were assessed by the Epworth Sleepiness Scale (ESS) to measure symptoms of EDS. Lumbar puncture was performed to measure and analyze Hcrt values in spinal fluid. Participants from the University of Rochester Medical Center also underwent tests to measure their sleep patterns in the laboratory. These sleep exams included a polysomnography (PSG) and a Multiple Sleep Latency Test (MSLT).

Results indicated that Hcrt-1 values and Hcrt splicing in DM1 were not different compared to controls. Levels of Hcrt-1 were also similar between patients with DM1 who reported varying degrees of EDS symptoms on the Epworth Sleepiness Scale (ESS). Both Hcrt-1 level and ESS score were not found to be associated with the size of CTG repeat expansion. There were abnormal results observed during the sleep studies. Patients with DM1 had a short sleep-onset REM. The results of these sleep exams and laboratory tests helped the authors conclude that excessive sleepiness in DM1 is different from the sleep disturbance that occurs in narcolepsy. More research is needed to determine the causes of excessive sleepiness in DM1 and the most appropriate therapies.

Chronic pain in DM and FSHD (2008)

Jensen MP, Hoffman AJ, Stoelb BL, Abresch RT, Carter GT, McDonald CM (2008). Chronic pain in persons with myotonic dystrophy and facioscapulohumeral dystrophy. Arch Phys Med Rehabil. 89(2):320-8.

"Results indicated that 60% of DM and 82% of FSHD patients reported pain."

Paper published by colleagues at the University of Washington and University of California, Davis.

These authors reported a high prevalence of pain in DM and FSHD patients. The paper highlights that 91% of the participants in this study were members of the Registry (235 of 257 total enrollees). The paper also highlights the eagerness of our population to participate in research, as 296 individuals contacted our colleagues at UC-Davis to participate, of which, 235 (79.4%) subjects completed and returned their questionnaires.

Their results indicated that 60% of DM and 82% of FSHD patients reported pain, with 23% of these respondents reporting that their pain was severe. Most patients reported having pain symptoms for over 10 years. The most common sites of pain were located in the lower back (74% of FSHD patients and 66% of DM patients) and legs (72% of FSHD patients and 64% DM patients). Average pain was greatest for those patients with assistive devices. Overall, pain was reported to have a moderate degree of interference with enjoyment of life (mobility, work, recreational activities).

Part of the author's conclusions indicated that there remains, "*too few options for pain relief of patients with DM and FSHD and chronic pain. There is a substantial need for the development to effective and long-lasting pain treatments that can be made easily available that have a few negative side effects.*"

These results and other information reported by Dr. Jensen and colleagues indicate that pain is a very significant symptom of DM and FSHD. More research and clinical guidelines are needed to identify treatments for pain.

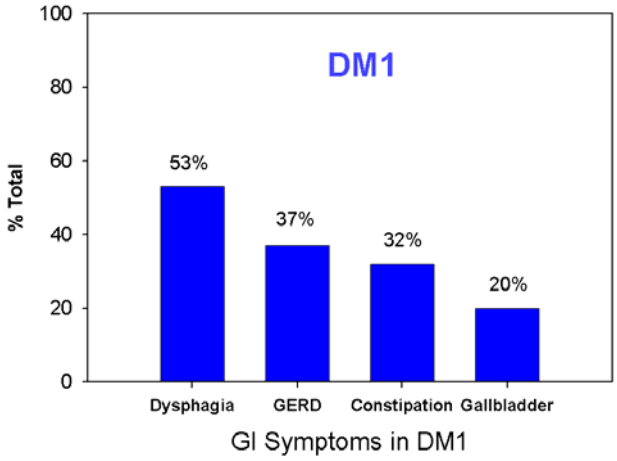
Gastrointestinal (GI) symptoms in DM

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 overall prevalence of symptoms and most frequently used treatments. Even less information is currently known about potential GI symptoms in DM2 patients. The lack of information suggests that these symptoms in DM2 patients are under-reported or absent compared to DM1.

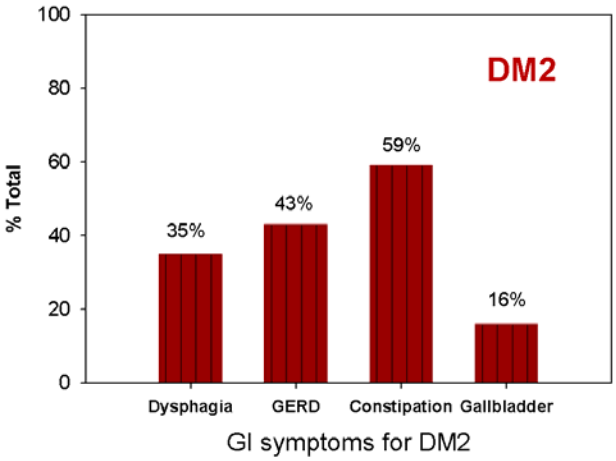
We analyzed GI symptoms reported by all DM1 and DM2 patients enrolled in the National Registry to learn more about these symptoms.

Results: Results presented in September 2008. GERD= Gastro-esophageal reflux disease; Dysphagia = trouble swallowing.

Presented by Registry staff at the Annual Meeting of the American Neurological Association , September 2008



GI Symptoms in DM1



GI symptoms for DM2

Conclusion

GI symptoms are common in DM1 and DM2 patients in the Registry and support results of a small Dutch study (Suominen, *Neuromuscular Disorders*, October 2007). We also report that GI symptoms may relate to greater weight and disease duration in DM1.

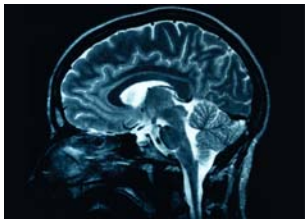
Further studies are necessary to help determine what causes GI symptoms in DM patients and how doctors can better manage symptoms. We hope to report these findings in an upcoming paper and will keep you updated as more information is available.

"GI symptoms are common in DM1 and DM2 patients enrolled in the Registry..."

Current studies recruiting Registry members

More detailed descriptions and contact information for the studies below are being sent by mail to all eligible Registry members.

We will provide more information to all members on our website and in our next newsletter as recruitment continues and when study results are available.



Example of a brain scan.

Cognitive function in DM

Study investigator: Dr. John Day at the University of Minnesota.

Study title: *Structural and functional central nervous system (CNS) changes in adults with myotonic dystrophy type 1 (DM1) and myotonic dystrophy type 2 (DM2).*

Dr. Day and his research team are trying to better characterize the cognitive changes that often occur in myotonic dystrophy patients.

The cognitive effects of DM are often diverse. Some patients may have mild cognitive symptoms or none at all. Other patients may have moderate to severe symptoms. Manifestations may include learning disabilities, excessive daytime sleepiness, mental retardation, decreased executive function, and psycho-social problems (depression, anxiety, anti-social behavior, etc). Investigators are trying to determine the underlying cause of these and other brain symptoms in DM1 and DM2.

Dr. John Day and his study team are recruiting 140 volunteers to participate in a study at the University of Minnesota. Patients will be asked to undergo a Magnetic Resonance Imaging (MRI) scan of the brain. Volunteers will be asked to complete intelligence tests, language tests, tests of memory and attention span, and tests of visual perception, spatial orientation, and abstract reasoning. Volunteers will also undergo a brief physical exam and will be asked to provide blood and skin samples.

"Knowing the gene responsible for FSHD is critical in the development of effective treatments."

Study of genetics in FSHD patients

Study Investigator: Dr. Rabi Tawil at the University of Rochester

Study Title: Cellular and Molecular Pathophysiology of Facioscapulohumeral Dystrophy (FSHD).

Dr. Tawil and his research team are trying to better understand the causes of FSHD. This study will attempt to determine which gene is affected in FSHD by comparing DNA results with clinical information. Studies such as this one are important, because knowing the gene responsible for FSHD is critical in the development of effective treatments.

130 volunteers will be recruited to participate in this study, 90 of which will travel to the University of Rochester. Dr. Tawil and his team are interested in examining eyes and muscle tissue because studies have shown that blood supply to these areas is affected in FSHD patients. Researchers do not fully understand these findings and whether reduced blood flow and perhaps other vascular or blood vessel problems contribute to the muscle wasting in FSHD. To investigate these ideas further, study participants will be asked to have a physical exam, an evaluation of their muscle strength, a needle muscle biopsy to obtain tissue for research, and an ophthalmologic examination, including a retinal vascular study.

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:

- better understand the natural course of aging with a disability (including those with MD);
- test the effectiveness of interventions in managing depression in people with disabilities as they age;
- enhance the employment experience of those with a physical disability;
- publish findings for people with disabilities, their family members, and their health care providers.

To aid in the goal of disseminating our findings, we plan to publish a quarterly informational column called "Aging Well with MD" through the National Registry. These columns will be posted on the website of the National Registry every 3-4 months. Additionally, all of the columns will be printed in the Registry's annual newsletter. Each column will focus on a specific topic of interest to those aging with MD and their family members, along with a description of the findings from our RRTC as they become available. The goal will be to provide information that is directly practical, informative, and based on the best science available.

When possible in our columns, we will summarize important new findings from the research literature as they are published, making them directly accessible to patients. But we also plan to include practical information that may be well known to health practitioners and the research community, but perhaps less well known to patients and their family members (for example, scientifically proven methods for helping improve sleep or manage pain). We strongly encourage input from our readers to help guide the content of our column. If you have a specific topic of interest relating to aging with MD that you would like to see highlighted in a future column, please send an e-mail to agerrtc@u.washington.edu.

"The purpose of this Center is to study the challenges faced by those aging with MD and other physical disabilities.."

Disclaimer

The summaries and recommendations that appear in this page are based on the expertise of the University of Washington's Aging Rehabilitation Research and Training Center and not directly from the National Registry or endorsed by NIH. The contents of this column were developed under a grant from the Department of Education, NIDRR grant number H133B080024. However, those contents do not necessarily represent the policy of the Department of Education, and you should not assume endorsement by the Federal Government.

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.

UNIVERSITY of ROCHESTER MEDICAL CENTER

National Institutes of Health
The Nation's Medical Research Agency