Volume 7

June 2010

National Registry of DM and FSHD Patients and Family Members

Introduction

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.

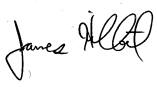
In the coming years, we anticipate exciting developments in the research and clinical care of DM and FSHD. Researchers have made significant advances in explaining the causes of symptoms of DM and FSHD, and understanding the cause of a symptom is a critical step towards treating it. Experimental treatments 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 of information for investigators and patients to identify the best measurements to use to prove that a treatment is having a beneficial effect.

As we prepare for these new therapies, the Registry needs to continue its growth and continue to provide a vital and unique resource for researchers to conduct studies. Such studies will allow us to better understand how to measure symptoms and how they change over time, and to better understand the issues important to patients and their families. Additionally, these studies will prepare researchers for potential clinical trials by determining which symptoms should be targeted for treatment and how much of a change would indicate that a treatment was effective.

Thank you again for your help in making the Registry a success and for your continued support in the coming years of the National Registry.

Best wishes for a happy and healthy summer!

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



James Hilbert, M.S.
Research Coordinator

Elizabeth Luebbe
Research Coordinator

Special points of interest:

- Enrolled over 1,600 members after 10 yrs!
- Presentations on FSHD clinical symptoms, early onset DM1, and diagnostic delay in DM2;
- Research paper on pain in DM1 and FSHD;
- Studies recruiting members of the Registry;
- Columns about "aging well" with muscular dystrophy

Inside this issue:

New Funding of the National Registry in September 2010

The National Registry has been "housed" or located at the University of Rochester since 2000 and funded by the National Institutes of Health (NIH). The NIH — *the Nation's Medical Research Agency* — includes 27 Institutes and Centers and is a component of the U.S. Department of Health and Human Services. It is the primary federal agency for conducting and supporting basic, clinical and translational medical research, and it investigates the causes, treatments, and cures for both common and rare diseases. NIH provides research funding through a variety of mechanisms, including grants, contracts, work study programs, and student loan repayment. The Registry has been funded by a *NIH contract* from September 2000 until Sept 2010.

After September 2010, the National Registry will partner with the University of Rochester Senator Wellstone Muscular Dystrophy Cooperative Research Centers (MDCRC) — more information on the MDCRC is on the next page. Funding of the University of Rochester MDCRC is sponsored by an *NIH grant*. Through this partnership, the Registry will remain at the University of Rochester, and we will maintain the stringent guidelines to protect your safety and confidentiality according to NIH guidelines. Such a partnership will help us to continue to provide families with new information and research opportunities related to DM and FSHD.

What does this mean for Registry members?

In summary, nothing will change for Registry members. Funding and oversight of the Registry will still be provided by NIH. The leaders and staff who you're familiar with, and who have worked on the Registry for years, will be maintained. Your information will be kept securely locked and protected in our database and remain protected by University and NIH guidelines. We currently have large cabinets that are "home" to all of your paper forms. These forms are secure and double locked in our hallways and maintained by other University safeguards.

The National Registry will maintain its current goals and procedures. As a member, you will continue to receive study notifications, annual updates, and newsletters from the Registry.

The transition will enable us to continue promoting the use and growth of the National Registry including educational opportunities about research studies in DM, FSHD, and other muscular dystrophies. Other opportunities exist to maintain sharing information about what types of new and exciting research is being done nationally and around the world and how these experimental treatments may lead to new drugs for DM and FSHD!

Where can I get more information?

If you have any questions, please feel free to call us toll-free at (888) 925-4302. We welcome your suggestions about the Registry through phone calls, letters, notes on your forms, or through email.

Please remember that your participation in the Registry is voluntary. You are free not to take part or to withdraw at any time, for whatever reason, without risking loss of present or future care you would otherwise expect to receive. In the event that you do withdraw from this study, the information you have already provided will be kept in a confidential manner. Please call our toll free number if you would like to withdraw from the Registry.

What about the short term and long term growth of the Registry?

We are very excited to maintain the strengths of the Registry and are hopeful that our growth will be sustained by additional funding beyond 2013. We will keep all our members updated on how the Registry may grow through future letters, updates to our website, and annual newsletters.

Please read more information on the National Registry's partnership with the University of Rochester Wellstone MDCRC on the next page!

SENATOR PAUL D. WELLSTONE MD COOPERATIVE RESEARCH CENTER

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.

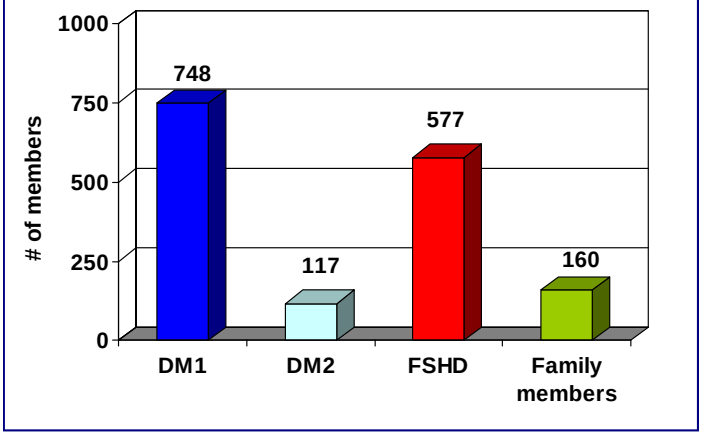
Project 2: "Experimental Therapy of Myotonic Dystrophy"

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.

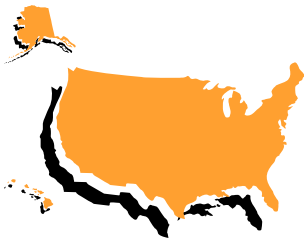
Enrollment updates of the Registry

Composition of the National Registry



To date, over 1,600 members have enrolled in the National Registry. This total represents 748 members with myotonic dystrophy type 1 (DM1), 117 with myotonic dystrophy type 2 (DM2), 577 with FSHD, and also 160 unaffected family members. **Our membership represents all 50 states!**

Members have assisted in the growth of the Registry to reach over 1,600 members by encouraging their affected and unaffected family members to join. In fact, 35% of Registry members have indicated that they have at least one family member enrolled. Our goal is to connect as many patients as possible with the research community and to hear from as many patients as possible with DM and FSHD. Every patient has a unique presentation of their disease and has an important individual or personal perspective on what symptoms have the biggest impact on their life. To gain a more complete and accurate picture of DM and FSHD, it's important that we recognize *all* of these presentations and perspectives. Therefore, we'd like to hear not only from all affected and unaffected family members who have already joined, but also from those not currently enrolled in the Registry. We encourage your help in inviting these family members to join.



Registry members continue to show their dedication to research by returning an annual update of their information. Eighty-six percent of Registry members have returned one or more annual updates! With this annual information from all of our members, we have one of the largest resources of patient-reported data on myotonic dystrophy and FSHD. The overall goals of the Registry are to:

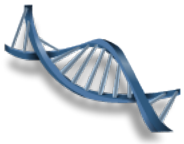
"To gain a more complete and accurate picture of DM and FSHD, it's important that we recognize all presentations and perspectives of DM and FSHD"

- **Build teamwork amongst doctors, investigators, and patients:** such teamwork helps us to share information about exciting opportunities and advances in DM and FSHD research and offers a great opportunity for doctors and investigators to learn from patients and hear about what symptoms are most important to them.

- **Collect and report clinical information:** the information that Registry members provide on questionnaires when enrolling into the Registry is important because it helps us to understand symptoms and their progression and management — **see pages 6-7.**

- **Recruit DM and FSHD patients into studies:** we help investigators recruit subjects to participate in clinical studies in research centers across the US, or in their home by completing questionnaires. Each study helps doctors, investigators, and others better understand the biology, progression, and other issues of DM and FSHD — **see pages 8-11.**

Updates on the biology of FSHD



The symptoms of FSHD are quite variable and can range from mild to severe. Weakness most often occurs in muscles of the face, upper back (scapular region), chest, upper arms, and ankles. Weakness can progress over time to affect the hip and trunk or stomach muscles. Other symptoms may include hearing loss, pain, and problems with the blood vessels of the retina.

DNA is made up of chemicals that provide "instructions" to make proteins and to carry out the functions of all life. These instructions or genes are passed on from generation to generation. DNA and proteins are tightly packaged together in chromosomes in cells of the body. Humans have 23 pairs of chromosomes and about 30,000 genes!

New research studies have provided more evidence on "how" FSHD occurs. Over 95% of individuals with FSHD have a form that researchers now refer to as FSHD type 1 (FSHD1). This form of FSHD results from the loss (deletion) of a specific segment of DNA at the tip of chromosome 4 (please see sidebar for information on genes and chromosomes!).

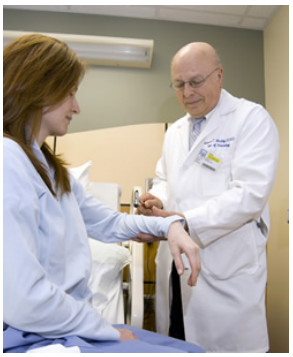
Over the past several years, scientists have found that in about 2% of individuals with FSHD, there is no deletion or loss of DNA on chromosome 4. For these individuals, a DNA test will show a negative result, suggesting that they do not have FSHD. When the sample is further analyzed with specialized research techniques, scientists will, instead, find a change in the chemical structure of the DNA in the same region of chromosome 4 that results in an unfurling of the DNA structure. This unraveling of DNA causes a second form of FSHD that is now referred to as "FSHD2." Both FSHD1 and FSHD2 appear to have identical clinical manifestations and scientists believe the disease-causing mechanism is the same in both, despite the different changes on chromosome 4. Research is now underway to determine how FSHD1 and 2 are connected and tests for FSHD2 are only available in research settings at the present time. If you have questions about your DNA results and whether further testing would be beneficial for you, please talk to your physician or genetic counselor.

What is the difference between DM1 and DM2?

Both DM1 and DM2 result from expansions of a certain part of DNA.

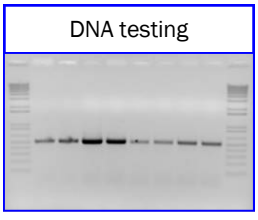
Both myotonic dystrophy type 1 (DM1) and type 2 (DM2) share many similar symptoms. Examples of shared symptoms include trouble relaxing muscles (myotonia), cataracts, and heart abnormalities. There are also many differences in symptoms between the two DM subtypes. DM1 is characterized by more weakness 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.

The gene defect for DM1 was discovered in 1992 and results from an expansion of a certain part of DNA. DNA carries "instructions" for a living organism to grow and function. DNA determines such things as hair color, eye color or other more complex traits.



DNA is made up of four chemicals represented by the letters, A, G, C, and T. These letters stand for the chemicals: adenine (A), guanine (G), cytosine (C), and thymine (T). The expanded piece of DNA in myotonic dystrophy contains CTG repeats.

The gene defect of DM2 was discovered in 2001. DM2 is caused by an expanded piece of DNA containing CCTG repeats.


DNA testing

Both of these pieces of abnormally expanded DNA in DM1 and DM2 cause problems in cells — the building blocks of the human body. The expanded repeats of DNA (CTG for DM1; CCTG for DM2) lead to changes in the cell that cause symptoms of DM.

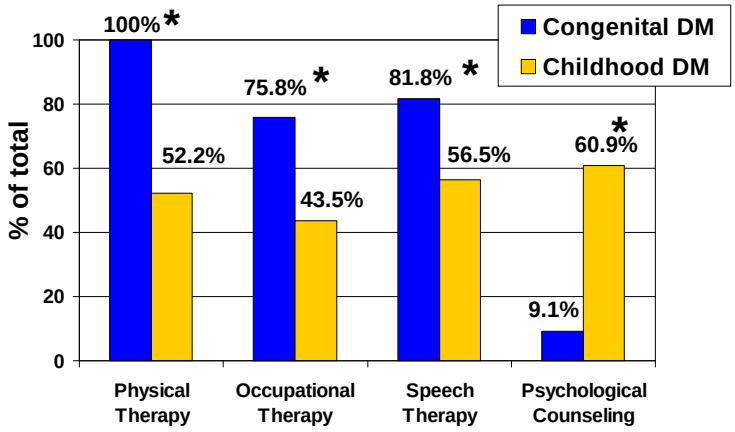
Clinical information reported by Registry members: Two presentation at the 7th International DM Consortium Meeting

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.

Use of Rehabilitation Therapies in Congenital and Childhood DM1



** Indicates significant difference between Congenital and Childhood DM1*

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 better characterize childhood-onset DM1 and to define issues that are unique to this group of patients.

Are there delays in diagnosing myotonic dystrophy type 2 (DM2) ?

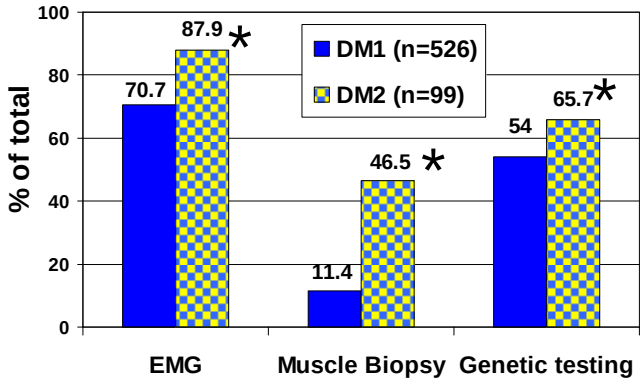
Limited information is known about possible delays that may occur in establishing the diagnosis of DM1 or DM2. Some researchers have shown that DM2 patients may be incorrectly diagnosed on their initial evaluation. For example, patients may be misdiagnosed with fibromyalgia (a muscle disease associated with pain and weakness).

We looked at information in the Registry to help us determine the potential delays in getting diagnosed in DM1 and DM2 patients. Information reported by our members indicated that

DM2 patients took an average of 15 yrs to be diagnosed (double the time compared to DM1).

Information in the Registry also showed that DM2 patients had undergone significantly more diagnostic procedures including genetic testing, EMG, and muscle biopsies compared to DM1.

Diagnostic Tests Received in DM1 and DM2



** Indicates significant difference between DM1 and DM2 ($p<0.05$).*

More education of care providers and patients is necessary to overcome the delay in diagnosis. Early and correct diagnosis of DM can improve disease management, preventative care, and getting proper physical therapy and other treatments.

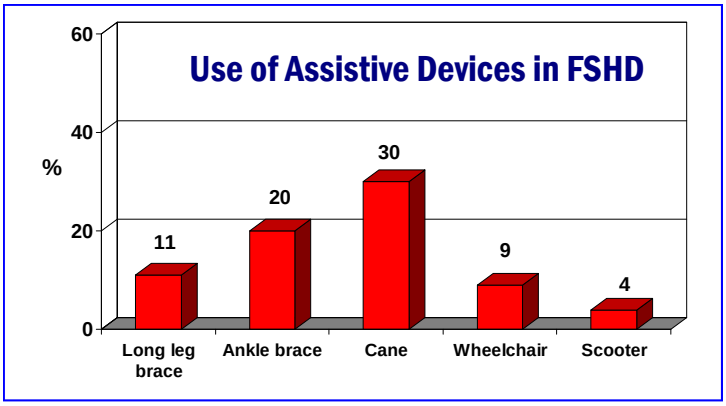
How else has my Registry information been used to study DM and FSHD?

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

Over the past year, we have mailed over 1,600 letters to help investigators recruit DM and FSHD members into their studies! While every member participates in the Registry by providing information on their symptoms as noted on the previous pages, contacting investigators to participate in studies is always voluntary! These studies often involve travel, and the investigators are often looking for only a select or small group of patients. We are hopeful that more studies will develop in the future and you will continue to participate!

More detailed descriptions and contact information for the studies below are being sent by mail to all eligible members of the Registry. We will also provide more information to all members about these studies on our website and future newsletters when results are available. Below are brief updates!

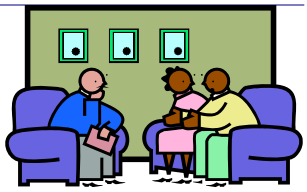
Survey of aging and muscular dystrophy

Study investigator: Dr. Mark Jensen, University of Washington (Seattle, WA); study supported by the National Institute for Disability Rehabilitation Research

Investigators are studying how aging affects individuals with muscular dystrophy and individuals with other physical disabilities. Research subjects will be asked to complete a series of surveys about their experience with pain, fatigue, sleep, participation in daily activities and employment, social support, and mood. This study is part of the University of Washington's recently funded Rehabilitation Research and Training Center.

Update from the study team: Researchers have enrolled 337 people into this study, and would like to thank everyone who took part in the study. Your participation helps us better understanding aging and how it relates to DM and FSHD! We are analyzing the results and are eager to report our findings to patients, doctors, and investigators.

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 research. Many thanks!

"Thank you!"

from the

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.

- 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!

Cognitive function in DM

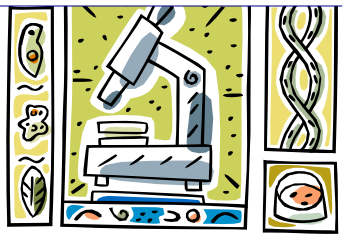
Study investigator: Dr. John Day, University of Minnesota (Minneapolis, MN); study supported by the National Institute of Neurological Disorders and Stroke (NINDS)

Researchers are studying central nervous system (CNS) changes that often occur in DM. The CNS consists of the brain and spinal cord, and it is responsible for "processing" signals for the entire body, both voluntary and involuntary.

CNS symptoms in DM are very diverse and range from none, mild, to severe symptoms. Examples include learning disabilities, excessive daytime sleepiness, decreased executive function, depression, and anxiety. Investigators 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 of memory and attention, 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

Update from the study team: Initial findings from this study show a dramatic loss of white matter integrity in MRI images in DM patients compared to unaffected volunteers (matched controls). Our neuropsychological data indicates there is relatively well preserved memory and language function in DM compared to changes seen in executive function and visual-spatial abilities. We are in the process of analyzing these data in childhood onset DM1, adult onset DM1, and DM2 subjects, and will re-evaluate the data longitudinally as subjects return for their second study visits in the upcoming years.

Collection of cell lines in DM



Study investigator:s Dr. Lisa Kalman, Center for Disease Control and Prevention (Atlanta, GA); Dr. Richard Moxley, University of Rochester (Rochester, NY) & Dr. Lorraine Toji, Coriell Cell Repositories (Camden, NJ); study supported by the CDC and NIH.

Investigators are collecting blood samples from patients with DM and family members to create cell lines which will be stored confidentially in the Coriell Cell Repository. These cell lines will provide an important resource for researchers studying DM and will also help ensure the quality of genetic testing for the DM community. Subjects will provide blood samples which will be stored anonymously and will be available for use in research, teaching, and as standards in clinical genetics laboratories.

Update from the study team: We are actively recruiting and greatly appreciate those who have expressed interest in the study so far. Letters announcing this study and asking for volunteers will be mailed to additional eligible Registry members in the coming months.

Study of disease progression in DM

Study investigator: Dr. Richard Moxley, University of Rochester (Rochester, NY); study supported by the National Institute of Neurological Disorders and Stroke (NINDS)

Investigators are studying the causes of muscle weakness and stiffness (myotonia) in DM. The study also aims to find the most effective way to measure symptoms of DM and to determine how symptoms change over time. This study will help prepare for future clinical trials by determining how to measure whether a potential treatment is effective.

Update from the study team: We have received an overwhelming response from Registry members interested in this study! Subjects have begun to travel to the University of Rochester for study visits, and we continue to enroll eligible subjects with DM1 and DM2. Thank you for your interest and participation in this important study!

FSH Society, Inc. Patient and Researcher Network Meeting July 30 - August 1, 2010, Las Vegas, Nevada

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
* How to Motivate Young People to Exercise
* Parents: Preparing for the Education and Work Life of your Children
* Maintaining a Good Diet and Good Nutrition
* The Expert Patient: Managing Dialog with Your Physician
* Advocacy and Disability Rights
* Dating and Forming Lifelong Relationships

The keynote address *"Developing and Delivering New Classes of Drugs to Improve Muscle Function and Health"* will be delivered by H.Q. Han, M.D., Ph.D., Scientific Director, Metabolic Disorders, Amgen, Inc., Thousand Oaks, CA

Distinguished clinicians and investigators in FSHD research are on the program, including:

- Joshua O. Benditt, M.D. Pulmonary and Critical Care Medicine, University of Washington Medical Center, Seattle, WA
- Leigh Ann Curl, M.D., Orthopedic Surgery, Sports Medicine, Knee and Shoulder Harbor Hospital, Baltimore, MD
- Melanie Ehrlich, Ph.D., Tulane Medical School, New Orleans, LA
- Charles P. Emerson, Jr., Ph.D., Boston Biomedical Research Institute, and BBRI Wellstone MD CRC, Watertown, MA
- Scott Q. Harper, Ph.D., The Ohio State University and Center for Gene Therapy, The Research Institute at Nationwide Children's Hospital, Columbus, OH
- Michael Kyba, Ph.D., University of Minnesota, Minneapolis, MN
- Craig M. McDonald, M.D., Ph.D., Rehabilitation Research and Training Center in Neuromuscular Diseases, University of California, Davis, CA
- Stephen J. Tapscott, M.D., Ph.D, Fred Hutchinson Cancer Research Center, Seattle, WA
- Rabi Tawil, M.D., Fields Center for FSHD & Neuromuscular Research and University of Rochester, Rochester, NY
- Kathryn Wagner, M.D., Ph.D., Center for Genetic Muscle Disorders, The Kennedy Krieger Institute, Baltimore, MD, and BBRI Wellstone MD CRC, Watertown, MA

For more information, please email 2010Meeting@fshsociety.org, or call the FSH Society at 617-658-7878. More information is also available on the FSH Society's website (www.fshsociety.org) which includes details on their meeting this year, plans and meetings for 2011, and current research activities and patient information.

Aging well with muscular dystrophy

In our last newsletter, we were excited to introduce "educational columns" written by colleagues at the University of Washington in Seattle, WA. The National Institute on Disability and Rehabilitation Research has recently funded these researchers to develop a Rehabilitation 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. Along with developing research studies, one of the main goals of this new Center is to publish findings and clinical care guidelines to people with disabilities, their family members, and their health care providers.

To help meet this goal, researchers from Washington have published quarterly informational columns called "Aging Well with Muscular Dystrophy." New columns are posted on the National Registry's website every 3 months. Columns currently available on the Registry website include the topics below:

- Overview of the RRTC and aging well with muscular dystrophy (March 2009)
- Sleep problems (June 2009)
- Physical and other factors that affect sleep (October 2009)
- Sleep problems and medications (January 2010)
- The burdens and management of pain (March 2010)

We would like to especially thank Dr. Mark Jensen and Kara McMullen for writing these columns! *The contents of these columns were developed under a grant from the Department of Education, National Institute on Disability and Rehabilitation Research (NIDRR); grant # H133B080024.*

UNIVERSITY of ROCHESTER MEDICAL CENTER

National Registry of DM and FSHD Patients and Family Members

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We'd like to thank everyone who has played a part in the growth and success of the National Registry. This of course includes our dedicated members: those that have been with us since the start-up of the Registry as well as those that have joined us along the way. All of the exciting information shared in this newsletter would not have been possible without your participation.

We'd also like to thank the many organizations that have recently promoted the Registry, particularly:

- Fields Center of FSHD and Neuromuscular Research
- FSH Society, Inc.
- Johns Hopkins University
- Muscular Dystrophy Association (MDA)
- Myotonic Dystrophy Assistance and Awareness Support Group (MDAASG)
- Myotonic Dystrophy Foundation (MDF)
- University of Iowa
- University of Washington

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