National Registry of DM and FSHD Patients and Family Members

Volume 8
March 2012

Dear members and friends,

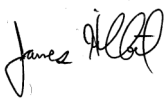
We hope this newsletter arrives to find you and your family in good health and excellent spirit. We are happy to report that Registry members like you remain committed to research and help advance knowledge of myotonic dystrophy (DM) and facioscapulohumeral muscular dystrophy (FSHD). Important points for discussion about the Registry and ways you help are highlighted throughout this newsletter and below:

- ***What is the importance of enrolling people of all "severities" into the Registry (page 2)?***
- ***Can your Registry information (re: medications, leg braces, surgeries, etc.) be used to improve the care of all patients (page 2-4)?***
- ***Are there exciting new insights in DM and FSHD that may lead to new drugs (page 5-6)? Are we prepared for new treatments (page 7)?***
- ***How can all our voices be heard (page 10-11)?***

The Registry helps give voice to the concerns and hopes of DM and FSHD patients and family members. The heart of the Registry is our 1,800 members. Each member brings unique stories and histories. Your voice is heard when you interact with researchers (on paper, on the phone, and in person) and when you answer questions from the Registry every year. All these answers "add up" and help researchers and care providers better understand and report your symptoms and how they change over time. And with each answer, and with each year, we and others around the world are better understanding and managing symptoms today and getting closer to developing therapeutic treatments for the future.

With much hope and perseverance, we remain steadfast to continue the growth of the Registry and to share a brighter and stronger future with you.

Warmest regards,



Richard Moxley, III, MD
Principal Investigator

James Hilbert, MS
Research Coordinator

Elizabeth Luebbe, MA
Research Coordinator

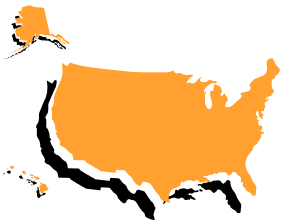
UNIVERSITY of ROCHESTER MEDICAL CENTER

Inside this issue:

Our Members

We continue to build momentum in the Registry. Throughout this growth, our goals remain the same to connect researchers and patients to advance research and to hear from you each year to learn more about your symptoms and the progression of DM and FSHD.

The Registry has enrolled 1760 members since enrollment began in November 2001. 818 individuals have enrolled with myotonic dystrophy type 1 (DM1), 137 with myotonic dystrophy type 2 (DM2), 632 with FSHD, and 173 unaffected family members. Patients from every state have enrolled in the Registry with highest representation in the South (29.4%), followed by the Northeast (25.0%), Midwest (24.7%), and West (20.9%).



1760 members from each and every state!!

Is anyone too young, too healthy, or too sick to join?

As we strive to reach more and more patients, we thank you for your continued support of the Registry. You have supported us by filling out your forms, telling your friends and family about the Registry, and sharing your experience and stories with us! Every person contributes to our success and guides us forward.

No one is too young or old, or too severely or mildly affected, to join the Registry. Infants and children to older adults and anyone in between are enrolled in the Registry and give us another piece of the puzzle in understanding DM and FSHD. *Your family members are eligible to enroll too.* If they're interested, we'd be happy to have them join. Without hearing from these individuals, we may be missing what's important to them, and have an incomplete picture of DM and FSHD.

Is it important to fill out the questionnaire I get every year?

Every year, we send you questionnaires to better understand how your symptoms progress, how your medications change, and how other medical problems may develop or improve. Members have sent back updates for up to 10 years! To date, average follow-up is 3.3 years in DM1, 3.1 years in DM2, and 4.0 years in FSHD. We would like to keep these return rates very high, so please keep up the great work!

Is my information important?

The answer to the question above is a resounding "yes!" By answering questions on your Registry forms about symptoms, assistive devices, and how your disease changes over time, we are able to analyze and report information about DM and FSHD to researchers, patients, and care providers. Such information from the Registry was recently published in the medical journal, *Contemporary Clinical Trials*!

"This information… may help doctors to develop clinical care guidelines and may help spur new research!"

The figure below shows an example of how we use your information to learn about DM and FSHD. This Figure summarizes the percentages of Registry members who reported use of assistive devices (e.g. cane or walker) for ambulation (like walking). Registry data indicate that muscle weakness will progress for about 5-10% of patients to require use of a new or different assistive device (like a wheelchair) after about 4 years. Wheelchair use for both short and long distances was reported by 6.3% of DM1, 8.3% of DM2, and 18.1% of FSHD members. This information, from one of the world's largest populations of DM and FSHD patients, may help doctors to develop clinical care guidelines and may help spur new research!

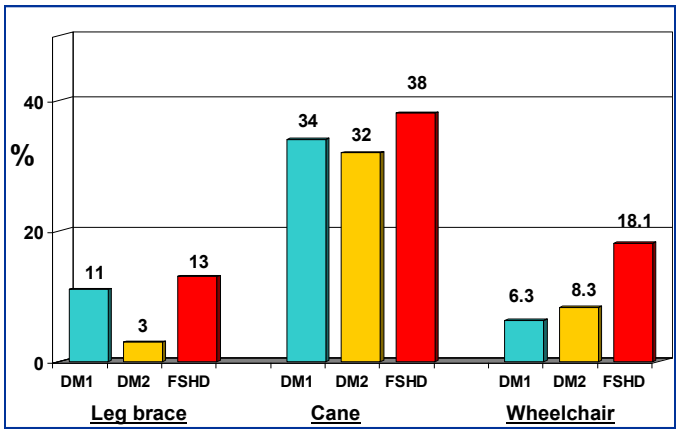
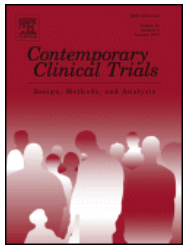


Figure 1: Please note these numbers are averages, and each member of the Registry has a unique combination and varying severities of symptoms of DM and FSHD.



Title: If you build a rare disease registry, will they enroll and will they use it? Methods and data from the National Registry;

Authors: Hilbert JE, Kissel JT, Luebbe EA, Martens WB, McDermott MP, Sanders DB, Tawil R, Thornton CA, Moxley RT 3rd; The Registry Scientific Advisory Committee.

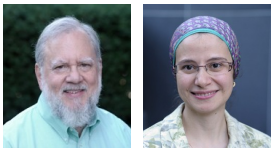
Contemporary Clinical trials, 2012 Mar;33(2):302-311

Potential risk for certain cancers in DM

The symptoms of DM are very diverse and can vary amongst family members. DM can affect the brain, eyes, stomach, heart, muscles, and other organs. **Each person is affected differently-** some patients have no or few symptoms while others have many different symptoms. Previous research from the Registry hinted that DM patients may also be at risk for certain cancers.

New research provides even more evidence. These results were recently published in the Journal of the American Medical Association (JAMA). The research studied DM subjects in Sweden and Denmark and showed that they may have an increased risk of cancers of the brain, ovary, colon, and the uterine lining known as the endometrium. The research was conducted by scientists at the National Cancer Institute, leaders of the National Registry at the University of Rochester, Denmark (Statens Serum Institut) and Sweden (Karolinska Institutet). ***These results are only preliminary. More research is needed*** with different populations (groups of patients) and studies are needed to determine the "biology" of why this may be true.

The authors encourage patients to discuss cancer screenings, symptoms, and risk factors with their primary care doctors and muscular dystrophy and cancer specialists. These results highlight the diverse nature of DM and how much more research and guidelines of care are needed!



Pictured from left to right; Drs. Greene and Gadalla from NCI.

Developing standards of care

Many patients share the experience of not always having their questions related to DM and FSHD adequately addressed. This can negatively impact patients' care and may be due primarily to a lack of information and resources in DM and FSHD. To help ensure that patients receive better care, development of a "standard of care" for DM and FSHD patients is needed. By taking part in the Registry, you help promote the development of these clinical care guidelines. For example, a meeting was held in 2010 to discuss "standards of care and the management of FSHD patients." De-identified (anonymous) information you provided to the Registry was presented at this meeting about the use of rehabilitation therapies (like physical therapy) and assistive devices (like wheelchairs). Information presented at the meeting was summarized and recently published in a medical journal for all care providers, researchers, and patients to use! Visit www.sciencedirect.com/science/article/pii/S0960896610001884 to read the article.

"To help ensure that patients receive better care, development of a "standard of care" for DM and FSHD patients is needed. By taking part in the Registry, you help promote the development of these clinical care guidelines."

Similar work is being done by researchers and care providers in DM. Recently the American Academy of Neurology (a professional group of over 25,000 doctors) has started to develop standards of care for DM. Such work involves reviewing all published information on DM, talking to expert doctors around the world, and comparing ideas. This work is ongoing and leaders hope to publish information this year or next. De-identified information you provide to the Registry will be part of these discussions!

Studies recruiting Registry members

The Registry has recently approved **9 studies** to use the Registry to recruit patients or to analyze anonymous data. Brief updates about these recent and ongoing studies are provided on pages **4-5 and 7-8!** These studies primarily focus on improving your care and preparing patients, family members, and care providers for new treatments.

Why haven't I heard about some of these studies?

Depending on the study design, researchers may only be recruiting patients with certain criteria. For example, researchers may only be studying patients within a certain age group, disease type, or weakness. If you didn't hear about some of the studies on the next pages, it could mean that the study was only looking at a particular group of patients or that study letters haven't been sent out yet. Also, if you recently joined the Registry or moved, we may have missed you. If you have any questions or concerns, please call us toll-free at 1-888-925-4302.

What matters most to you?

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

Pictured:
Dr. Heatwole

Investigators are studying the symptoms that are often most important to DM and FSHD patients, such as pain, fatigue, gastro-intestinal problems, and weakness. Investigators of this study are sending surveys for members of the Registry to provide insights into how their disease impacts their life. Information from this study will be used to help develop disease-specific quality-of-life questionnaires to be used in future clinical trials. The Registry team has sent these surveys to over 1,100 members in 2010 and early 2011!

Update from the study team: Thank you very much for your help in completing and returning these surveys. About 60% of you have replied! This information is invaluable!! It will be one of the largest surveys of DM1, DM2, and FSHD ever reported! We are continuing to analyze these surveys and hope to report the results in medical journals and to you soon. Your guidance will help us better track what symptoms matter most to you and how we can better measures these symptoms in future research studies.

Helping teach pharmacists about muscular dystrophy

Study investigators: Dr. Richard T. Moxley, III, University of Rochester (Rochester, NY) and Dr. Amy L. Parkhill, Wegmans School of Pharmacy, St. John Fisher College (Rochester, NY)

"Knowing which medications are used in DM and FSHD patients is a 'first step' approach to better understand the breadth of symptoms and hopefully better manage care."

Limited information is known about what types of medications patients with DM and FSHD use and how these medications may relate to symptoms. In this study, researchers and pharmacy students are analyzing anonymous data in the Registry related to medications and clinical symptoms. There are over 5,000 medications reported by DM and FSHD members. The researchers are categorizing these medications and comparing usage amongst DM and FSHD patients and information reported by the general public. Opportunities exist to present this information at national pharmacy meetings, in medical journals, and to ultimately help educate pharmacy students who often have limited knowledge of muscular dystrophies.

This study also highlights important ways you can help! It's very important to report your prescriptions correctly and report all over-the-counter medications, supplements, or drugs. Knowing which medications are used in DM and FSHD patients is a "first step" approach to better understand the breadth of symptoms and hopefully better manage care.

Cognitive function in DM

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

Pictured:
Dr. Day

Investigators are studying how the brain is affected in DM. Examples are excessive daytime sleepiness and problems with attention, memory, and motivation. Researchers are recruiting 140 volunteers to undergo brain scans, tests of memory and attention, brief physical exams, and to provide blood and skin samples. The study has enrolled ~75 people with DM1, DM2, and congenital DM1 and hope to recruit additional subjects in 2012. Additional recruitment letters will be sent soon to eligible members of the Registry! Preliminary results of the study show small structural changes in the brain in certain DM subjects compared to those without DM. Other preliminary results indicate strong memory and language skills but reduced motivation, attention, and visual learning. The exact location of these changes in the brain, their potential implications, and other information are still being analyzed.

The study team is very thankful for the support of the Registry and for the many patients who have participated. **From the team:** it has been great meeting many of you! We look forward to working together to help study cognitive changes in DM and to one day better manage and hopefully treat these symptoms.

Progress in FSHD Research in the Fields Center assisted by National Registry

Synergy between the NIH supported National Registry and the Fields Center partners in Rochester, Seattle, and the Netherlands, has helped to support important advances that will help patients and researchers. Below is a summary of some of the advances that have occurred during the past year.

Patients, Families and Scientists: Partnering to Achieve Important Advances in FSHD

The Fields Center for FSHD & Neuromuscular Research is pleased to reflect upon the year since our last update as it gives us another opportunity to thank the members of the National Registry for their active involvement. Thanks to you, scientists are beginning to make exciting breakthroughs in FSHD research!

In the last year, members of the National Registry have continued to volunteer for FSHD studies available through the University of Rochester. Overwhelmingly, volunteers have given us permission to share de-identified information as well as blood, muscle and skin samples with our colleagues worldwide. Because of these generous samples, scientists have made several important discoveries. Highlights of accomplishments the past year include:

- This research led to publication of the unifying theory as to the mechanism causing FSHD and identification of the probable gene involved (DUX4). Now, scientists have a potential target to go after to develop treatments for FSHD.

- Further research confirmed that protein from the DUX4 gene, normally active only in germline cells (sperms and eggs) continues to be expressed in muscle of individuals with FSHD whereas it is turned off in individuals who do not have the FSHD genetic defect. This information provides support for the idea that one option for treatment will be to try to suppress the expression of this gene.

- Publication of research showing two genetically distinct forms of FSHD now known as FSHD1 and FSHD2. Both FSHD1 and FSHD2 have identical physical features on examination but look different on DNA testing. Whereas individuals with FSHD1 have a deletion on chromosome 4, individuals with FSHD2 (less than 5% of all FSHD) do not have a deletion. Yet individuals with FSHD2 have loosening of the DNA structure on chromosome 4 similar to that seen in FSHD1 and in both, the turning on of the normally silent DUX4 gene is thought to be the culprit.

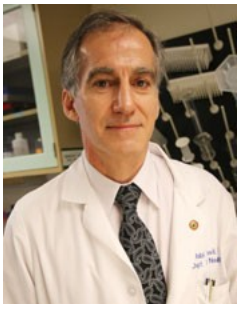
- Fields Center partners at the University of Rochester and Leiden University Medical Center (The Netherlands) hosted two international meetings to propose "best practice" standards for clinical care and laboratory testing methods. The data generated through your research visits and ongoing laboratory testing is helping to shape the practice patterns of clinicians and researchers worldwide.

Thank you for continuing to support our efforts. We are keenly aware that, without your contributions, we would not be able to make inroads into this disease. Together, we are making a difference. We look forward to continuing to work with you to move forward toward development of treatment options. For more information about the Fields Center, please contact us:

- By phone: 585.275.7680
- By email: FieldsCenter@urmc.rochester.edu
- Through our website: www.FieldsCenter.org

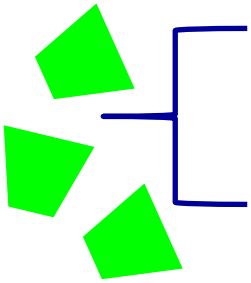
We look forward to hearing from you!

"Thanks to you, scientists are beginning to make exciting breakthroughs in FSHD research!"

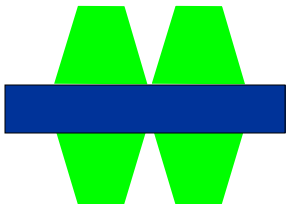


Pictured:
Dr. Rabi Tawil

Healthy cell



Proteins "stuck" in DM1 cell



Biology and experimental therapies for DM

DM results from an abnormal expansion of a certain part of DNA (deoxyribonucleic acid). DNA carries instructions for a living organism to grow and function. It is a major component of the cell nucleus. DNA determines such things as hair color, eye color or other more complex traits. RNA (ribonucleic acid) acts like a "messenger" to read the instructions on DNA and to guide the formation of cells and structures of the body. RNA regulatory proteins in the nucleus act like the "foreman" on the job. They guide proper function of reading the blueprint (see schematic diagram on the left).

In DM, these nuclear regulatory proteins are "stuck" to abnormally expanded RNA that is retained in the nucleus. This "sequestration" prevents their proper function to read the blueprints of the cell. Without receiving proper instructions, many of the diverse symptoms in DM can occur (see left). Researchers around the world are making important and exciting steps to reverse the toxic effects of the abnormal RNA in the nucleus by studying animal models of DM. These researchers are cautiously optimistic that they can develop new treatments to remove the toxic RNA or to release the RNA regulatory proteins from being stuck so they can do their normal job.

Much work needs to be done to make sure such treatments will be safe and effective for humans. We remain hopeful that they will. We'll keep you updated on research progress and look forward to you helping us and other investigators bring these treatments into human studies and hopefully to all patients!

Overview of clinical studies

The recent advances in DM and FSHD research have generated much enthusiasm, hope, and anticipation for effective treatments in the future. This wave of excitement has also generated questions, confusion, and misinformation regarding when, how, and for whom treatments may become available. Like you, we want treatments to be available as soon as possible. However, drug development is a long, complex process that requires many phases of research to determine if a potential treatment is both safe and effective for people. Each phase (outlined below) is necessary to bring experimental treatments to patients. Each phase can take 1-3 years and can require up to tens of millions of dollars of funding.

- Preclinical- studies in the laboratory with cell models and animals
- Phase 1- studies with a small number of patients to determine safety and drug dosage
- Phase 2- studies with more patients to determine safety and effectiveness
- Phase 3- studies in a large population to determine effectiveness

Photo of the Clinical Research Center, courtesy of University of Rochester

The good news: we're very excited and hopeful because we're moving in the right direction. You can read more background information about research studies on our website (dystrophyregistry.org) or the NIH database of clinical trials (clinicaltrials.gov).

What's being done now?

Current studies are underway to help "pave the way" for such treatments trials (next page). Examples include trying to determine the exact progression of DM and FSHD and to analyze which procedures are most accurate, reliable, and tolerable to patients to measure the small and large changes in disease symptoms (changes in weakness, blood levels of certain proteins, etc) that occur during short or longer intervals of time. Such information is crucial so that when treatments are available, study measures can help determine if new drugs and treatments are working.

Are we prepared for new treatments in DM?

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

Pictured: Dr. Moxley

Researchers involved in the study "Pathogenesis and Progression in Myotonic Dystrophy (DM)" are investigating the causes of muscle weakness, stiffness (myotonia), and the progression of other DM symptoms. **From the study team:** We're enrolling 80 DM1 and 20 DM2 patients to have an initial study visit and follow-up study visits after 1 and 3 years. The study aims to find the most effective ways to measure symptoms of DM and to determine how symptoms change over time. Are certain muscle tests (strength and walking distance) more important to use in clinical studies and are they able to detect large or subtle differences in the progression of DM? Answers to these questions are important to guide the clinical care of patients and to determine the effectiveness of future experimental therapies!

Recruitment into the study is nearly complete. Approximately 1/3 of the subjects in the study have been recruited through the Registry. We would like to thank everyone that has been interested and those who have been able to participate in this study. Your help is very much appreciated!!

Study investigator: Dr. Tetsuo Ashizawa, University of Florida (Gainesville, FL); study supported by the Marigold Foundation

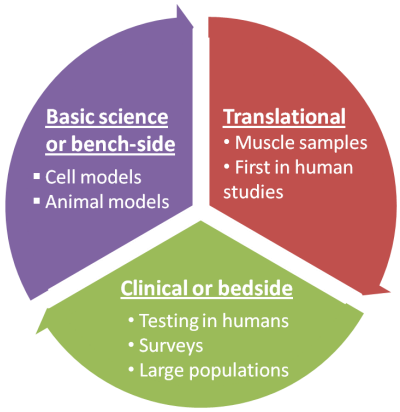
The purpose of the Myotonic Dystrophy Biomarkers Discovery Initiative (DMBDI) is to study factors that influence the biology of how DM occurs and progresses. For example, researchers are looking for certain factors that could be used in a laboratory test (like RNA molecular markers in muscle biopsies) that indicate how DM may be changing over time. The Marigold Foundation (a Canadian charity) sponsored this study and it was conducted at 3 national and 3 international sites. The University of Florida was one of these clinical sites. They thank all of the individuals (and there were many) who responded to the Registry's mailing. Only a very few patients were enrolled (due to a shorter than expected enrollment window); but, the study team was highly impressed by the volume of responses received in just a few weeks. The study team sends a big thank you and best regards to the Registry team and to all of the members of the Registry for their effort and support!!

How can information in the Registry help prepare for treatments trials in FSHD?

Study investigator: Dr. Jeffrey Statland, University of Rochester (Rochester, NY)

Investigators at the University of Rochester are analyzing changes in disease burden and functional ability from de-identified (anonymous) data reported by FSHD members in the National Registry. **From the study team:** We're hoping to learn more about how FSHD affects patients' lives over time by looking at changes reported by Registry members year after year. In particular, we're looking at changes in use of assistive devices, mobility, and symptoms associated with FSHD. We continue to analyze this data and will report results to you when available. We hope to use this information to help design functional assessment tools to be used in future clinical trials and to gain a better understanding of the natural history of FSHD.

How else does the Registry help move research from "bench to bedside"?



Bench research: Collection of cell lines in DM

Pictured from left to right; Drs. Toji and Kalman

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

The purpose of this study is to collect blood samples from patients with DM and family members to create cell lines. These samples will be used anonymously by investigators and health care professionals across the world for research, teaching, and as standards in clinical genetics laboratories. For example, these cells will be used as "quality controls" to help confirm that newly diagnosed patients have DM and to ensure the continued quality and accuracy of DM genetic testing.

About 100 DM members from the Registry were contacted to participate in this study. These members have low and high numbers of DM repeats. The interest in this study was amazing! "Blood collection" kits were mailed to 40 members to have their blood drawn by their local doctor. Of the 26 samples returned so far and processed for creation of cell lines, 20 were successful, 1 is still in progress and 5 may be unsuccessful. DNA from the earliest submissions has been sent for molecular confirmation. These cell lines are now being analyzed to confirm the number of DM repeats. Thanks to the cooperation of Registry members these new cell lines will soon become available for research and for reference materials for clinical diagnostic laboratories worldwide!

Bedside research: Aging well with dystrophy

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

Pictures courtesy of University of Washington School of Medicine

From the study team: The University of Washington's Rehabilitation Research and Training Center (RRTC) would like to extend thanks and gratitude to everyone who has completed and who continue to complete our surveys. The goal of these surveys is to measure how aging affects certain symptoms and quality of life for people with muscular dystrophy and other physical disabilities. Researchers hope to better understand the frequency, course, and impact of symptoms such as pain, fatigue, and depression on aging with a disability. Additionally, collecting data on participation in employment, communication, social support and other facets of life that are affected by aging with a physical disability will increase our understanding of these important issues. To date, below is a summary of persons with muscular dystrophy (MD) who participated in our surveys:

- Year 1 Survey (2009 – 2010): 340 participants/ 382 expressed interest (89%)
- Year 2 Survey (2010 – 2011): 189 participants/ 203 expressed interest (93%) *This survey was limited to individuals ages 45 – 65.

Your responses to the surveys are wonderful and clearly show a dedication to participating in research about aging with MD. We're preparing to send out our Year 3 Survey. We have published one paper on fatigue, and we're writing two additional papers -- one on pain and fatigue and the other on employment issues. We will share the findings from these papers shortly. We can be reached at agerrtc@uw.edu or 1-866-932-6395 if you have any questions about our research. Thanks again for your participation!

Do we know enough about DM and FSHD in children?

We often hear from patients that their motivation to get involved in research is to help their children someday. Parents are eager to do whatever they can to improve the quality of life for their kids who have DM or FSHD. We're eager to do whatever we can too.

The first step in this process is to make sure we know enough about DM and FSHD in children. Children, who have symptoms from birth or early childhood, often have different symptoms and unique needs compared to individuals who start having symptoms as an adult. We need to better understand these symptoms and their progression in order to improve care for children and ensure that they'll be able to receive treatments when available. Additionally, we need to make sure that we have sensitive and reliable ways to measure symptoms in children so that we can ensure that potential treatments are safe and effective for children. Most of all, we need to better understand what's most important to them and their families.

"Parents are eager to do whatever they can to improve the quality of life for their kids who have DM or FSHD. We're eager to do whatever we can too".

How is the Registry helping researchers study children?

Clinical Features of Infantile FSHD

Study investigator: Dr. Jean Mah, University of Calgary (Alberta, Canada); study supported by the FSH Society and Muscular Dystrophy Canada

This study aims to better understand the clinical characteristics of infantile FSHD. Although it makes up only about 5-10% of the total FSHD population, understanding infantile FSHD is important. These patients are often more severely affected and have unique symptoms, including seizures as well as problems with cognition, hearing, and vision. Dr. Mah hopes to gain information about the impact of infantile FSHD on quality of life,, determine how genetics may be associated with symptoms, and to determine ways to measure the "natural history" or progression, of FSHD in children.

Pictured: Dr. Mah

The study team is enrolling 50 subjects with infantile FSHD at several national and international research centers. The Registry hopes to send letters to all eligible members (or parents of eligible minors) with more information in the next few weeks.

Early-Onset Hearing Loss in FSHD

Study investigator: Dr. Katherine Mathews, University of Iowa (Iowa City, IA)

Researchers in this study are interested in learning more about patients with FSHD who experience hearing loss in early childhood. Little information is available regarding hearing loss for these children. The hearing problems often require the use of hearing aids and can become disabling. The goals of this study are to learn more about this subset of patients, including any associated clinical and genetic characteristics and understand those factors contributing to the progression of hearing loss over time.

Photo courtesy University of Iowa

Researchers plan to mail a survey to eligible Registry members (or parents of eligible minors) and to also look at anonymous information in the Registry for members with FSHD who have reported use of hearing aids.

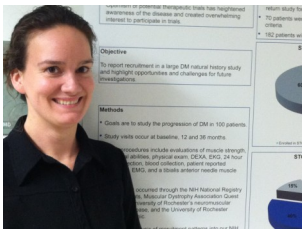
Quality of Life in Early-Onset DM

Study investigator: Dr. Nicholas Johnson, University of Rochester (Rochester, NY)

This study aims to determine the symptoms that have the greatest impact on quality of life for patients with myotonic dystrophy who have symptoms from birth (congenital myotonic dystrophy) or early childhood (juvenile myotonic dystrophy). These patients have unique symptoms and their disease symptoms need to be compared to those with adult-onset DM. Little information is available regarding what is most important to them and their families.

Dr. Johnson plans to mail a disease-specific, quality of life questionnaire to all eligible Registry members to determine what may be the most important symptoms to them and their parents. The research team plans to use this information to help finalize their questionnaire for use in future studies and clinical trials.

Patient perspectives: a survey about participating in research studies



Pictured: Ms. Dekdebrun

Study investigator: Dr. Richard Moxley and Jeanne Dekdebrun, MS; University of Rochester (Rochester, NY)

The different types of research studies and the reasons why certain patients are selected to participate can be confusing. Therefore, helping patients better understand the process of research studies is important. The goals of this survey are to better understand patient perspectives so that researchers may improve recruitment and enrollment into studies and to better understand how researchers can utilize each patient's willingness to help prepare for future treatment trials.

Advocating for patients with FSHD: the FSH Society

The FSH Society, a not-for-profit organization based out of Watertown, MA, has been a key leader for advocacy and support in FSHD for many years. According to the organization, their mission is a "cause without borders," with active roles in patient education and empowerment, networking, fundraising, and in the promotion and funding of basic, translational, and clinical research in FSHD. All of their efforts are aimed at finding treatments and a cure for FSHD.

How can I learn more or become involved?

If you're not already familiar with the FSH Society, please visit their website (www.fshsociety.org) for more information and resources for patients, family members, and care providers.

Upcoming Event

The FSH Society sponsors a biennial meeting dedicated to promoting interaction and networking between patients, clinicians, and researchers. Patients and research experts in FSHD from all around the world will be at the meeting. Highlights of this year's meeting are lectures and "round table" discussions with experts in FSHD, updates on clinical and research advances in the field, and educational information on a variety of topics. Examples include discussions on ways to improve quality of life, "managing dialogues with physicians," exercise, family planning, and interacting with other patients and advocates. Brief details about the meeting are provided below, with greater details and registration information available at: www.fshsociety.org/pages/comMeeting.html. **Meeting registration required by June 1, 2012.**

"Highlights… include discussions on ways to improve quality of life, 'managing dialogues with physicians,' exercise, family planning, and interacting with other patients and advocates."

The Biennial FSHD International Patient and Researcher Network Meeting

Atlanta Marriott Marquis

265 Peachtree Center Avenue NE

Atlanta, GA 30303

June 30, 2012 – July 1, 2012

**Hotel registration required by June 7, 2012*

Call 800-266-9432 or 506-474-2009 for hotel reservations

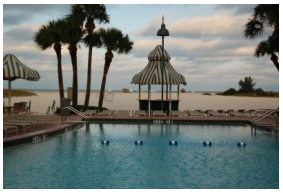
Photo courtesy of the Atlanta Marriott Marquis

Meeting of the "minds & hearts" about DM research and patient care

8th meeting of the International Myotonic Dystrophy Consortium (IDMC-8)

A large, international conference was held in December 2011 that brought together many of the "best and brightest" scientists and clinicians studying DM. This conference meets every other year. This time it was hosted by the University of Florida and held in Clearwater, FL. The main goals of the IDMC meetings are to bring researchers all around the world together to learn from each other and present information and findings from their work in DM.

The meeting included over 200 researchers from more than 20 countries, each working closely with each other, discussing "hot topics" and charting a common path ahead. More information about the conference sponsors, its program, pictures, and goals of IDMC can be found online at http://www.idmc.org/idmc8.html.



Clearwater Beach photo courtesy of Dr. Ranum, University of Florida

2nd Annual EMPOWER 2011 Family Conference

The Myotonic Dystrophy Foundation (MDF) led and helped sponsor a family conference in conjunction with the IDMC meeting and University of Florida. The conference was energetic, inspiring, and educational. It brought together over 400 family members, who were from 36 states and eight countries! The weekend included many presentations, question and answer sessions, and many discussions. Topics included the latest updates on research, patient care, and information on quality of life and daily living issues. The program of the conference, its sponsors, and videos of the conference talks are available online! http://www.myotonic.org/community/events/empower-2011/program/.

"Topics included the latest updates on research, patient care, and information on quality of life and daily living issues….videos of the conference talks are available online!"

Highlights of the Registry at conference

Your participation in the Registry was highlighted throughout the weekend by meeting many of you (!), encouraging more patients to join, presenting information on research studies you participated in, and by presenting data that each of you have completed on your Registry forms! Examples are listed below!

Biomarkers in DM by Katharine Hagerman , PhD, and the team from the University of Rochester. "Biomarkers" refers to signs of how our bodies react to injury or disease. Katharine measured what genes or DNA signals through the RNA it induces are "more or less active" in DM. This study analyzed muscle tissues donated from patients in the disease progression study highlighted on page 7! The researchers found interesting changes that occur to certain genes and RNA in severe and mildly weakened muscles. For this work, Katharine won an award for best clinical presentation (top picture, right) at the conference!

Gastrointestinal (GI) symptoms in DM by Jim Hilbert, MS, and the team from the University of Rochester. This study found that GI symptoms were very common in DM members of the Registry (77% of DM1 and 80% of DM2). Common problems included trouble swallowing, constipation, acid reflux, and gall bladder problems. The researchers also analyzed changes over time, and found that these symptoms progressed slowly. This information highlights the importance of GI symptoms in DM, and the need for more studies to develop better clinical care guidelines, understand biological causes, and design future treatments. For this work, Jim Hilbert (bottom picture, right) won a "best clinical poster" award at the conference!

Photos courtesy of Dr. Ranum, University of Florida

Highlights of other presentations at IDMC that used the Registry for recruitment or anonymous data can be found on our website (dystrophyregistry.org)!

National Registry of Myotonic Dystrophy & FSHD Patients and Family Members

Some members of our team: Ms. Luebbe, Mr. Martens, Ms. Eastwood, Mr. Hilbert & Dr. Moxley

Appreciation to all our members and collaborators!

Thank you for your continued support in the Registry – from filling out your forms, telling your friends and family about the Registry, and for sharing your experience and stories with us! Every person contributes to our success and guides us forward! We look forward to learning from each other and continuing on our "journey" together.

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

- Fields Center of FSHD and Neuromuscular Research
- Friends of FSH Research
- FSH Society, Inc.
- Muscular Dystrophy Association (MDA)
- Myotonic Dystrophy Foundation (MDF)
- University of Iowa
- University of Washington, Seattle

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