UNIVERSITY of ROCHESTER

MELIORA
Ever Better

National Registry of Myotonic Dystrophy & FSHD Patients and Family Members

November 2017

Dear Registry members,

Over the past 17 years, you have made the National Registry one of the largest and most diverse research communities of myotonic dystrophy (DM) and facioscapulohumeral muscular dystrophy (FSHD) patients!

The Registry now has enrolled more than 2,500 members and continues to grow. To say people have shared with us their "blood, sweat, and tears" is not an exaggeration. Members like you have shared so much about themselves and their families with us, donated blood samples in Registry-based studies, and undergone extensive muscle strength and function and other testing during research visits.

Your help to participate in the Registry and other research has led to new insights about DM and FSHD. It has also helped build the necessary tools (e.g., biomarkers and validated questionnaires) to accelerate the development of potential new treatments. This newsletter highlights important ways that you helped advance research over the past year and summarizes updates on other Registry activities.

Thank you for your commitment to learning more about DM and FSHD, to finding a cure, and to making a difference for your family and generations to come.

Inside this issue:

The University of Rochester's motto, *Meliora,* is a Latin word that means "ever better." We strive to be ever better in supporting research to ultimately improve the lives of patients with DM and FSHD.

Pictured: A conference for local patients, families, researchers, and clinicians at the University of Rochester.

Happy Retirement, Dr. Moxley

This past July marks the end of an era for the neuromuscular disease community with the much deserved retirement of Dr. Moxley from the University of Rochester.

Dr. Moxley is a world leader in neuromuscular research and has cared for patients and led research for over 43 years! Many of you interacted with him over the years as a patient in our clinic, a participant in studies in our clinical research center, or at research meetings and patient events. His limitless energy and enthusiasm are contagious and his commitment to patients speaks volumes about his character. Among notable accomplishments in the neuromuscular field, Dr. Moxley has:

- Helped discover and characterize myotonic dystrophy type 2 (DM2) in 1994 with Dr. Kenneth Ricker and other clinicians and researchers.
- Helped establish the University of Rochester as one of six nationwide centers of excellence through the NIH funded Senator Wellstone Muscular Dystrophy Specialized Research Center (MDCRC).
- Founded and led this Registry since 2000. It continues to grow and is one of the largest resources for clinical research and longitudinal data in DM and FSHD.
- Spearheaded numerous research studies for DM, including clinical trials and studies investigating DM symptoms as well as the mechanisms for how DM occurs and progresses over time. These studies and collaborations have led to knowledge about the multi-system complications of DM (such as myotonia, gastrointestinal symptoms, and insulin resistance), and use of mexiletine as an anti-myotonia treatment.
- Partnered with national and international researchers, government (e.g., NIH, FDA), pharmaceutical companies, patient advocacy groups, and patients and families to advance research and improve patient care.

Dr. Moxley's accomplishments can't fully describe his influence on the lives of his patients. Please read thoughtful notes from a few of Dr. Moxley's clinic and research patients to the right.

Same leadership and experienced Registry team

The Registry will continue to grow and facilitate research in DM and FSHD under the leadership of Drs. Rabi Tawil (pictured), Charles Thornton, and Michael McDermott. All three are international leaders in neuromuscular disease and have facilitated the growth of the Registry since its inception in 2000. Our Registry coordinators have been very fortunate to meet and interact with many of you – with Jim Hilbert and Liz Luebbe facilitating the Registry and clinical studies for the past 13 and 9 years, respectively.

"Once in a great while we are fortunate enough in life to meet someone that I call a finger of God. Dr. Moxley is certainly that person. His endless devotion to trying to help the patients and families with this horrific disease is irreplaceable. His name is now part of a very special history that has laid the groundwork for more incredible people to work from. We all love him, and wish him all the best. He will be sorely missed."

"I thoroughly enjoyed my visits with Dr. Moxley. He was always patient in answering all my questions. He continuously thanked me for being part of the study when I should've been thanking him for his time and devotion to the study of myotonic dystrophy. I am grateful for my time at the University of Rochester and wish to thank him now for the years he dedicated trying to understand this disease, its causes and treatments. "

Since I have been diagnosed I have been in touch with so many medical professionals but I have never experienced such great professional care as I had at the University of Rochester. Dr. Moxley and his "dream team" are absolutely amazing people who care deeply about their patients. My dear Dr. Moxley, your advice, recommendations, and most of all your encouragement have been helping me to fight my disease and stay positive. I and my family are grateful to you and Liz for all your support and care. All the best in the next chapter of your life! With deep appreciation and respect.

"If I am being honest, I did not look forward to the shots or overnight stays while I was involved in the clinical research facility. But, I really looked forward to the many times that Dr. Moxley came to see me. All of your patients will miss your smile, your calm and compassionate demeanor and how you always said thank you for being here. You are a real gentleman and a credit to your profession. You deserve this time to relax and enjoy yourself. I will never forget your kindness. "

Broad spectrum of research for DM and FSHD

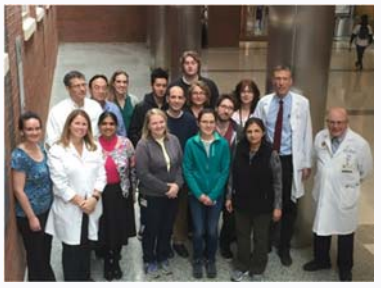
Many important steps are needed along the path toward developing and approving new drugs, including studying blood and tissues samples, recruiting patients who meet eligibility criteria (e.g., having genetic or DNA testing), refining experimental therapies, and learning more about symptoms of the disease. Below are a few ways that you've helped the National Registry drive national and international research in the past year! We will continue to contact eligible Registry members about a couple of these studies as well as new studies over the coming year.

Bio-resources for FSHD research and clinical trials (BIOACT): The University of Rochester is conducting a study which aims to help identify biomarkers in blood and tissue which may be good "targets" for experimental treatments for FSHD. The researchers are enrolling people with FSHD1 and FSHD2, as well as unaffected individuals. This study will continue to recruit eligible Registry members.

Genetic testing and outreach for FSHD: The University of Rochester and Leiden University Medical Center (Netherlands) are conducting a study to confirm the genetic status of Registry members with suspected FSHD. Genetic (or DNA) testing by a blood draw can determine whether a patient has FSHD1, FSHD2, or neither. Clinical trials for FSHD often require patients to have had genetically confirmed FSHD to participate. This study will be enrolling about 40 eligible Registry members per year over the next 5 years.

Experimental drug for DM1: Ionis Pharmaceuticals led a study to test an experimental drug in DM1 (an antisense oligonucleotide designed to get rid of toxic RNA in cells). The study was completed and enrolled 48 patients at 7 sites in the US. Results showed that the drug was safe and well tolerated in the form and dosages used in the study. Unfortunately, the study results did not show significant changes in many other measures, such as, strength and myotonia. However, insights from this trial and technological advances are helping the drug company develop a new drug that may improve potency and delivery to specific muscles.

Gastrointestinal (GI) manifestations in DM1 and DM2: We recently published a paper that describes GI manifestations in a large number of Registry members with DM1 and DM2. This paper was published in the journal, *Neurology*. Our data showed that 79% (n=721/913) of DM1 and 77% (n=139/180) of DM2 members reported a history of one or more GI symptoms. Both patients with DM1 and DM2 had a similar, high frequency of trouble swallowing (55% in DM1 and 29% in DM2), constipation (33% DM1 and 53% DM2), and high use of medications indicated for gastroesophageal reflux disease. In addition, about 20% of patients had their gallbladder removed, on average, about 10 years earlier than the general population.



More research is needed to better understand why these GI symptoms occur, how they impact health related quality of life, and how best to treat symptoms now and with future experimental therapies. Such data highlights the importance of your help to report DM symptoms that are under-studied and often have a big impact on your life.

More updates available online and via email: The studies above are only a small subset of important research being done with help from Registry members like you. Future updates online (dystrophyregistry.org) and via email will include results from a study of depression and medication use in Registry members; an update on a clinical trial for mexiletine use in DM1; new information about FSHD research through aTyr Pharma and Acceleron Pharma; and the development of care considerations for DM1, DM2, and FSHD!

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Get Connected with Us

The National Registry
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Phone (toll-free): 1-888-925-4302
Email: dystrophy_registry@urmc.rochester.edu

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Receive email updates

Receive email updates from us to learn about study results and to complete research questionnaires.

- If you've provided your email address to us, you should have received emails from us over the past year. If you haven't, but would like to, please contact us.
- You can opt to receive these updates through the postal mail if you prefer or if you don't have computer access.

Participate in research

Information will be mailed soon about opportunities to participate in clinical studies of DM and FSHD!

Visit our website

Visit our website (www.dystrophyregistry.org) for more information including research studies, press releases, 35+ published papers, newsletters, and more!

Thank you for your ongoing support of the Registry and research in DM and FSHD.