National Registry of Myotonic Dystrophy and Facioscapulohumeral Dystrophy Patients and Family Members

Celebrating 14 Years of the National Registry!

For 14 years, you have helped us collect information about the symptoms of myotonic dystrophy (DM) and facioscapulohumeral muscular dystrophy (FSHD). Thank you for sharing your stories and insights with us, participating in studies, and completing updates on your health each year. We continue to learn from members like you every day.

Many members have been a part of our Registry team since September 2000, while many others are just learning about their disease and are new to the Registry. Every member helps to make our Registry one of the largest, most well defined, and longstanding resources for DM and FSHD research. With you and researchers, we are building a stronger community to better understand symptoms and develop new treatments. 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.

"Learn, Discover, Heal, Create – And Make the World Ever Better"
We are inspired by how your participation in the Registry meshes with our University of Rochester's mission statement to "Learn, Discover, Heal, and Create." Your participation in the Registry and studies through the Registry has allowed the research community to:

- **Learn** more about the spectrum and severity of symptoms in DM and FSHD. Registry data has shown that:
  - Over a period of 6 years, 24% of FSHD members started using a wheelchair for at least part of the day. This risk is influenced by age and disease severity *(Publication #17*)*;
  - DM patients had to undergo repeated evaluations for over a decade (on average) to obtain their correct diagnosis *(Publication #19*)*;
  - There are new insights about skin and other tumors in DM *(Publications #6, 10, 15, & 21*)*.

- **Discover** the biological mechanisms of DM and FSHD, which symptoms may matter most to patients, and ways to help care providers better manage symptoms. Registry members:
  - Donated blood samples to help create a resource to improve clinical genetic testing in DM *(Publication #18*)*;
  - Provided evidence that Coats syndrome (eye disorder) occurs in FSHD almost exclusively in individuals with very large deletions *(Publication #16*)*.

- **Heal** symptoms in a mouse model of DM and learn more about how to target new drugs for FSHD. Registry members have helped make progress in bringing new treatments to patients by:
  - Donating blood and muscle samples to help researchers better understand what causes DM and FSHD and to help learn how symptoms may respond to future experimental therapies (*Press Releases**).

- **Create** new studies, networks, and partnerships with patients and researchers. The success of our Registry has led to:
  - Meetings and discussions with researchers about developing international registries for DM and FSHD in Australia, Canada, Germany, Italy, Netherlands, Peru, United Kingdom, and United States (*Publication #12**);
  - Collaboration with a Myotonic Dystrophy Clinical Research Network for patients to work closely with researchers and other stakeholders to become "trial ready" to test new treatments in patients.

*****Please see corresponding publications and materials on our website (www.dystrophyregistry.org) for more information.

The findings above are only a few of those from the Registry that have been published in research journals and presented around the world! **Learn more on the next page** or visit our website for a complete list of the Registry's impact on the research community**.**

Building upon partnerships with patients and researchers – and preparing for the future

Our growing community

The University of Rochester (home of the National Registry) was established as one of the first Senator Paul D. Wellstone Muscular Dystrophy Cooperative Research Centers (MDCRC) in 2003. These Centers are funded by the National Institutes of Health (NIH), which is part of the US government and has "the largest source of funding for medical research in the world." The NIH funds 6 Centers in a five year funding cycle to establish centers of excellence in muscular dystrophy. These Centers aim to work together to identify the causes of muscular dystrophy, measure disease progression, train young investigators, and develop new treatments. The University of Rochester was renewed as an MDCRC in 2008 and 2013.

The infrastructure and support of the MDCRC and other funders have led to the recent creation of a Myotonic Dystrophy Clinical Research Network (DM CRN). The DM CRN is led by the University of Rochester and consists of four other sites across the country: Ohio State University, Stanford University, University of Kansas, and the University of Florida. We are hopeful that an FSHD clinical research network can be developed quickly as well.

Our work together

The Registry represents 2,153 members and continues to grow! It also represents one of the most diverse communities of DM and FSHD patients with members from every state and from all ages and walks of life. The Registry continues to facilitate research by assisting investigators in recruiting patients into clinical studies. The Registry has facilitated 19 studies in the past 5 years! These studies are led by 15 different investigators at 11 different research sites. Haven't received a letter in a while? More information and updates are coming to you soon (see back page). The Registry also facilitates research by collecting data to learn important information about the progression and impact of DM and FSHD over time. We cannot learn more about how symptoms progress without your dedication and help in providing updates on your health each year.

Our impact

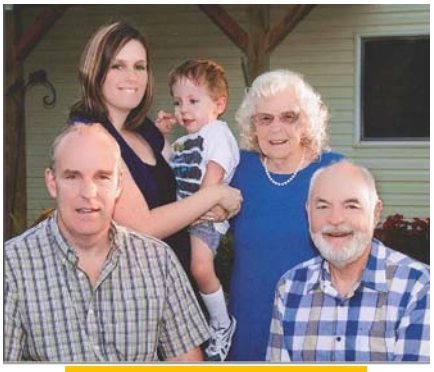
Your partnership with the Registry and other researchers has resulted in several influential presentations and publications. Visit our website for a complete list of these 23 publications. Below are select papers from the past 3 years:

- **Pain location and intensity impacts function in persons with DM and FSHD**
  University of Washington, published in: Am J Phys Med Rehabil; 2013 Nov 16.
- **Symptom Burden in Persons with DM and FSHD**
  University of Washington, published in: Muscle Nerve; 2014 Jan 11.
- **Correlates of tumor development in patients with DM**
  National Cancer Institute; published in: PLoS One; 2013 Nov 13;8(11).
- **Risk of functional impairment in facioscapulohumeral muscular dystrophy**
  University of Rochester; published in: Muscle Nerve; 2013 Jul 19.
- **High Prevalence of Gastrointestinal Symptoms with Progression over 5 years in DM1 and DM2 Patients**
  University of Rochester; received best poster presentation at an international research conference; 2011.

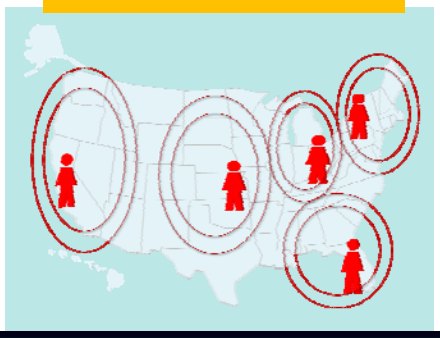
Our past, present, and future projects

The collage on the next page is a snapshot of many of our goals, publications, and ongoing research projects. Exciting projects ahead include surveys on medication use and pain, more frequent newsletters, clinical studies of DM and FSHD, and reports about the information you provide us every year.

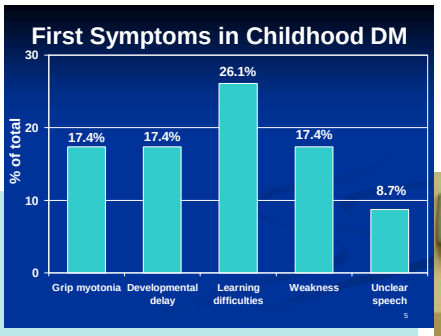
Teamwork

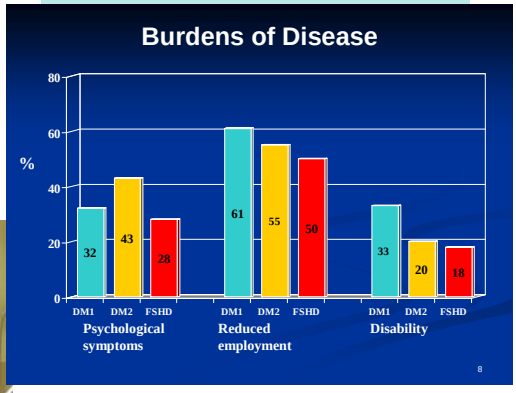


Patients Nationwide



Families





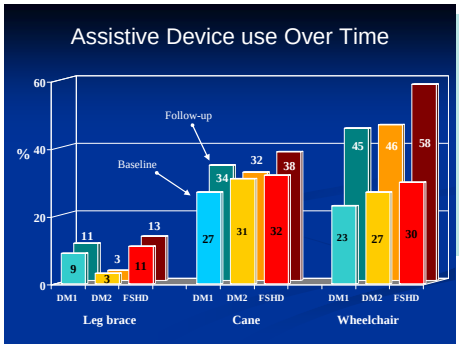
Research Papers

Presentations

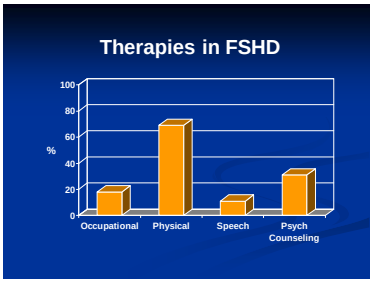
UNIVERSITY of ROCHESTER MEDICAL CENTER

Clinical Studies

National Registry of Myotonic Dystrophy and FSHD Patients and Family Members



14 Years and Growing



International Collaboration

NIAMS

NATIONAL INSTITUTES OF HEALTH

NINDS

UNIVERSITY of ROCHESTER MEDICAL CENTER

MEDICINE of THE HIGHEST ORDER

Stay better connected with us!

Follow us on Facebook!

Learn more about research progress through the Registry, read links to new, reputable sources of information about DM and FSHD, and get to know our team better! **To find us, search for "National Registry of Myotonic Dystrophy and FSHD."**

Receive email updates!

Receive email updates from us to learn about study results and how your participation is advancing research in DM and FSHD.

- If you've provided your email address to the Registry, you'll receive an email update soon. You are free to unsubscribe if you don't wish to receive these emails.
- You can opt to receive these updates through the postal mail if you prefer or if you don't have computer access.

Pictured above: Study coordinator Liz Luebbe discusses research findings with a patient during a clinical trial. One example of how the Registry connects patients with research teams!

Participate in new research!

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

Visit our website!

Visit our website (www.dystrophyregistry.org) for more information including research studies, press releases, 20+ published papers, newsletters, and more!

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

Call to learn more and have family members join!

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