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

Our newsletter comes with an exciting update that the Registry has been refunded for the next five years with support from the **National Institutes of Health (NIH)**. The Registry continues to grow and currently contains over 575 DM and 450 FSHD patients and 115 unaffected family members. Several important research projects are being developed that would not be possible without your support.

A few of our current and future research projects include:

- Presenting Registry information to physicians, researchers, and government officials to describe the symptoms of DM and FSHD and to help explain the need for more research.
- Measuring the short and long-term progression of DM and FSHD symptoms.
- Developing a research project to better measure how the burdens of muscle disease affect quality of life and daily activities. New types of questionnaires may make it easier and quicker to measure pain, fatigue, and other symptoms common to DM and FSHD patients.
- Supporting cell biology research to increase our understanding of muscular dystrophy, ultimately leading to the development of potential treatments.

NIH and other researchers are optimistic about the progress of the Registry, and we hope to build upon this success. In the months and years ahead, several opportunities exist to better track and potentially help manage the symptoms of DM and FSHD using the resources of the Registry.

We hope you share this excitement with us, and we send our best regards for the warmer spring and summer months ahead. Thank you again for your support and dedication.

Sincerely,

Richard T. Moxley, III, MD
Principal Investigator

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Research Updates

Researchers have given many presentations about the National Registry this past year. These presentations were given at research conferences, government sponsored health and research workshops, and meetings for patient support groups. A description of some of these presentations is listed below:

Presentations from the leaders and staff of the Registry

- Presented information at the NIH sponsored Burden of Muscle Disease Workshop in January 2005. Several research experts attended this conference including specialists in medical economics, physical therapy, psychology, muscular dystrophy, and cell biology. This presentation illustrated that the burdens of DM and FSHD are more than just physical and can affect employment, mood and sleep. The additional support that patients often need such as medications, canes, wheelchairs and therapies were also discussed.

- Presented information about the burdens of DM and ways to better measure patient reported symptoms at a research conference sponsored by the International Myotonic Dystrophy Consortium. Researchers and patients from all around the world attended this conference in Quebec City, in October, 2005 to learn about new advances and research about DM.

- Presented a research poster at the American Academy of Neurology meeting in April, 2006. This meeting is “one of the world's largest gatherings of neurology professionals, bringing together more than 10,000 neurologists and neuroscientists.” The poster described the short term progression of DM and FSHD symptoms and how researchers can use the resources of the Registry to conduct more studies.

Presentations from investigators using the Registry

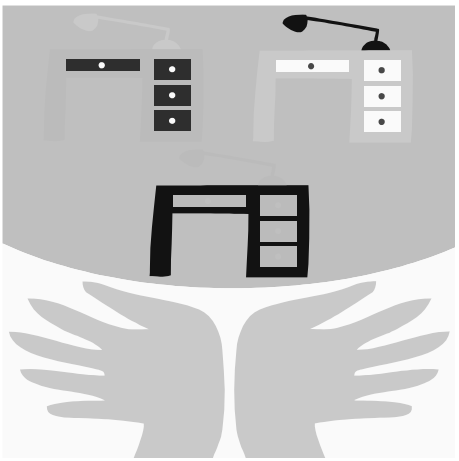
- Dr. Emma Ciafaloni from the University of Rochester Medical Center has presented information about the effects of excessive sleepiness in Myotonic Dystrophy at several research conferences. She has presented information at the American Academy of Neurology, Associated Professional Sleep Societies, and the International Myotonic Dystrophy Consortium. Her research indicates that excessive sleepiness is highly prevalent in DM patients but the reason or biological mechanisms for this sleepiness remain unknown. Future research may help discover the reasons and lead to new treatments.

- Dr. Emma Ciafaloni has also presented about the course and outcome in pregnancy from women with FSHD at the American Academy of Neurology Conference. Her research found that pregnancy and birth outcomes were generally favorable in this group of women with FSHD. However, the rate of certain obstetrical issues such as increased number of cesarean sections supports the need for additional research.

Registry Research Papers involving all Members of the Registry

We are currently writing several research papers about the Registry. Our first paper will describe the structure of the Registry. This paper is being written to:

- Help educate primary care doctors and specialists about DM and FSHD.
- Recruit more scientists to develop research projects to study DM and FSHD.
- Recruit more patients to join our Registry.
- Help researchers develop other disease registries.



Other Registry papers will examine the symptoms and progression of DM and FSHD. These research paper will include information about:

- Muscle weakness and the muscles of the body most affected.
- Muscle and other types of pain.
- The number of patients that use canes, leg braces, wheelchairs and other aids.
- Other health problems like high blood pressure, eye diseases, breathing difficulty, depression, and diabetes.
- The use of various therapies like physical, genetic, and occupational therapy.

The reports will use anonymous (un-named) information about every member of the Registry. Your enrollment in the Registry has been vital to this research.

Research Projects Recruiting Registry Patients

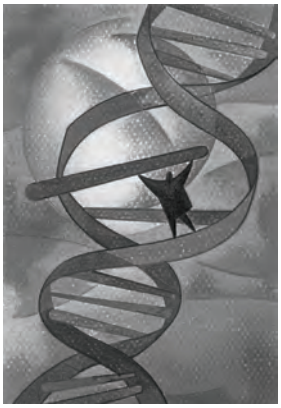
Dr. Craig McDonald from the University of California at Davis continues to study **The Quality of Life in Persons with Disabilities**. Dr. McDonald is seeking volunteers to complete a survey about their quality of life and to participate in several follow-up surveys. The main purpose of this study is to measure the cause and effects of chronic pain in persons with muscular dystrophy and other chronic diseases. He is also studying **Health and Wellness** and is recruiting patients to measure their exercise and wellness behavior.

Thank you for your support and assistance.

Website Updates

The Registry website has been upgraded with several new features improving the site for members, researchers, and health care professionals. The improved layout and organization of the website will allow for easier navigation and location of information. We have also added new sections to the website that provide educational resources and research updates.

The website's education section aims to address many of the common questions and concerns that members have about their illness. Our first installment is an introduction to genetic counseling and the genetic components of FSHD and DM. The guide describes an initial appointment with a genetic counselor and offers suggestions on information you should gather for your visit. A simple description of how FSHD and DM are inherited and the genetic testing process are also provided. In addition, we have supplied contact information for genetics counselors through out the United States, including counselors that specialize in muscular dystrophy.



We are also pleased to provide research updates for a number of projects that Registry members have participated in. This includes presentations we have given describing the structure and progress of the Registry. The website will provide brief project descriptions, journal articles and samples of poster presentations.

We encourage all of our members to visit the Registry website at www.dystrophyregistry.org. We will continue to keep you updated about any changes and will provide research updates as they become available.

New Email Policy

In response to recently enacted rules designed to maintain the privacy of your confidential medical information, the University of Rochester Medical Center has implemented a policy concerning the use of email as a communication tool between healthcare providers and patients. This policy has a direct impact on the National Registry as well.

From this point on, if you wish to exchange information with the Registry via email, you will need to sign a consent form that gives us permission to do so. This form, the Patient Email Consent Form must be signed by both you and the Registry staff. The form is an acknowledgement of the risks, responsibilities and the type of information that is appropriate to be relayed via email.

This form is available by contacting the Registry staff or for download from our website. If you want to use email to communicate with the Registry, please complete this form and return it to the Registry office. Or you can fax a copy of the signed form to the Registry office. We will return a signed copy of this form to you for your files. If you have any questions or concerns regarding this policy, please do not hesitate to contact us.

Annual Updates

Annual Update Forms are sent to all members of the Registry every year to study the progression or development of DM and FSHD symptoms. Your response to these annual updates has been very helpful. Many other research scientists are impressed with our highly motivated and dedicated patients! We have been collecting information for over 3 years and over 78% of Registry patients return these forms.

Annual updates will help us determine how quickly muscle or other medical problems develop. By studying this information, we and other researchers will have a better understanding of the biology of DM and FSHD. This increased understanding will help scientists develop better treatments and ultimately help prevent some of these symptoms. The information we have collected has been used in numerous presentations to illustrate the need for additional research in this area.

How can enrolled Registry members continue to help?

- continue to ***return your annual updates*** so that we can keep our database current. Your current information is vital for our research.

- notify the ***National Registry staff of any changes in your address or telephone numbers***. We would hate to "lose" you because we lack current contact information.

- ***watch for notices of research studies*** that are recruiting for volunteers.

- ***help recruit other patients*** to join the Registry by providing our contact information to your affected and unaffected family members and people in support groups. We would be happy to talk with them about joining the Registry.

- ***hand-out Registry flyers*** (see insert) to family members, doctors, or friends in DM or FSHD patient support groups.

Please feel free to make photo-copies of these flyers or let us know if you would need extra copies to hand-out. We believe our current members are an excellent way to help us recruit more patients - every additional member of the Registry helps us better study the full range of symptoms in DM and FSHD and promote future research.