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

Registry News!!

As all of us embark upon the New Year, and as we at the Registry prepare our application to the National Institutes of Health (NIH) to continue funding of the National Registry, we have feelings of hope, expectation and excitement. Over the next 2 to 3 years, as part of the new activities of the Registry, we hope to embark on Registry-sponsored studies that will involve the entire population of the Registry. For example, one of the studies we hope to undertake will use computer-based questionnaires developed by researchers in a large NIH funded project. These questionnaires deal with fatigue, pain, depression, and burden of care - problems that patients with DM and FSHD know all too well. We expect that the results will have immediate application and improve our ability to understand and measure the impact of these difficult problems in daily life. We are optimistic and excited that the results will allow us to improve the routine care of patients with DM and FSHD before we attain the ultimate goal of curing these illnesses. Look for a future announcement about this new study and about our plans for the Registry in the coming years. Have a wonderful New Year, and may 2005 bring each of you great satisfaction and hope.

Sincerely,

Richard T. Moxley III, MD
Principal Investigator

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Recruitment Update

After 3 years of recruitment, we have enrolled over 870 members into the Registry. We now have large amounts of information about the diseases. In an effort to look at the characteristics of the patients/members of the Registry, the Registry staff collected and analyzed anonymous information provided by our members. ***Data from every member of the Registry was used for this project!***

We are continuing to analyze this data and are preparing detailed scientific papers for submission to various journals within the next year. These articles will promote the Registry, help recruit researchers, and help further educate the scientific community. We sincerely appreciate your willingness to provide your information, as your input is the basis of the data we report to our colleagues in the medical community.

We have included examples of anonymous data we evaluated for your reference:

What is the average age of Registry members?

The average age of members affected with DM is 45 years. The average age of members affected with FSHD is 50 years.

How many males and females are in the Registry?

The Registry has 56% females and 44% males for DM. For FSHD, females comprise 53% and males represent 47% of the total population.

How did members discover the Registry?

Approximately 20% of DM and FSHD members were referred to the Registry by the Muscular Dystrophy Association. Other referrals included the internet (15%), doctors (10%), and support groups (15% for FSHD).

Where are the members located?

The Registry members are located in almost every state. We have members with DM living in 48 states and members with FSHD in 46 states.

The table below represents the geographic location of Registry members.

Region	DM (%)	FSHD (%)
Northeast	32	28
Midwest	24	26
South	27	26
West	17	19

How many applications have been sent? How many people are enrolled?

	DM	FSHD
Applications sent	915	845
Applications returned	583 (64%)	538 (64%)
Enrolled subjects	**465**	**409**
Affected adults	409	348
Affected child	23	8
Unaffected family	33	53

Besides muscle problems, what are some other medical issues affecting Registry members?

Problem	DM (%)	FSHD (%)
Acid reflux	36	24
Constipation	36	22
Gallbladder	18	6
Heart disease	18	13
Pneumonia	24	16

What other information can be presented?

The Registry has collected genetic (DNA) information from almost half of its members. DNA is the genetic material responsible for such things as eye color, hair color, and other more complex traits. This information is not required for entry into the Registry, but it can help researchers anonymously study various medical problems of these rare diseases. Other information that can be reported includes muscle problems, the use of assistive devices (braces, wheelchairs, etc), medications, and employment characteristics.

How can enrolled Registry members continue to help?

- We encourage you to ***offer our contact information*** to your affected and unaffected family members and members of your support groups. We would be happy to talk with them about joining the Registry.
- Please continue to ***return your annual updates*** so that we can keep our database current. This database is critical for "anonymous" research studies – your current information is vital for this research.
- Please 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!
- Please ***watch for notices of research studies*** that are recruiting for volunteers.

Research Updates

To date, the Scientific Advisory Committee of the National Registry has approved ten research studies. When a study is approved, the Registry searches the database for members who appear to meet the qualifying criteria set by the researcher. The Registry sends notices to those members, announcing the study and inviting them to call the researcher to volunteer if they are interested.

Examples of approved studies include:

- **The Course and Outcome of Pregnancy and Delivery in Women with FSH Dystrophy** (Emma Ciafaloni, MD. University of Rochester [NY]). Announcements were mailed to 143 women of childbearing age who have FSHD. Dr. Ciafaloni is looking for women to complete several surveys, asking information about their pregnancy, delivery and post-partum experiences.

- **The Quality of Life in Persons with Disabilities** (Craig McDonald, MD. University of California, Davis). Dr. McDonald was seeking volunteers to complete a survey about their quality of life and to participate in several follow-up surveys. The Registry contacted 454 members with DM and FSHD to announce the study.

- **The Pathophysiology of Hypersomnolence in Myotonic Dystrophy:** (Emma Ciafaloni, MD. University of Rochester [NY]). Announcements were sent to 210 Registry members who had FSHD, asking them to participate in a survey as control subjects to compare to the results obtained from subjects with Myotonic Dystrophy.

Feedback from these researchers has been quite positive! They are impressed by the response received from individuals seeking to participate! Thank you!

Recruitment for other studies is pending. Please watch your mail for other opportunities and volunteer if you are able. Many gains in research are possible when people collaborate with researchers to provide information, samples and personal insight. Your assistance is valuable!

Website Updates

The Registry website was updated in April 2004 to assist both patients and researchers. Since the upgrade, the number of "sessions" on the site has increased by 64% compared to last year. Patients can now review the goals and privacy regulations of the Registry, download forms and mailing labels, and get answers to frequently asked questions. The improved website also helps researchers by explaining the Registry and providing directions on how they can apply to use the Registry resources for their studies.

We encourage everyone to visit our website if you have internet access. Our internet address is located at www.dystrophyregistry.org.

For those without internet access who would like to view the website, we encourage you to use public computers. These computers can be found at libraries, open sessions at public high schools and universities, coffee shops and other stores, and by borrowing a friend or relative's computer.

We are constantly looking at ways to enhance and expand the Registry website. In the near future, we will explore ways to include links to various medical references, provide descriptions of active research, and possibly share anonymous data with researchers. *We are interested in your feedback about the website*. It will be used to help us determine how best to modify and maintain our site. Would you please take a moment to complete the brief survey inserted in this newsletter? Please let us know your thoughts – if you have any ideas and ways to improve the Registry website, please include these with your response. If you have feedback at a later date, you can email us at **dystrophy_registry@urmc.rochester.edu**

We will review all suggestions carefully! We will keep you posted about changes as they become available. Thank you for your input and support!

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Membership Survey regarding the National Registry's Website:

www.dystrophyregistry.org

1. Have you visited the Registry's website? ☐ Yes ☐ No

2. If you answered "Yes" to Question 1, do you check the website periodically for updates? ☐ Yes ☐ No

3. Do you have any suggestions for improving the site? ☐ Yes ☐ No

If you prefer to speak with us personally, the National Registry Staff can be reached (toll free) at 888.925.4302.

When you have finished this survey, please remove this page from the newsletter and return it to us in the postage paid envelope.

Thank you for your input!