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

Dear Registry Applicant,

Thank you for your interest in the **National Registry of Myotonic Dystrophy and Facioscapulohumeral Muscular Dystrophy Patients and Family Members**! The Registry was established through a contract with the National Institutes of Health to link people with Myotonic Dystrophy and FSHD with researchers who are studying these rare diseases. At this time, we are registering individuals with FSHD and DM, as well as unaffected family members. Therefore, we invite you to encourage other family members to apply.

In this packet, you will find the forms necessary for your **child** to apply to the Registry. To apply, please complete the following steps:

1. There are two identical *Permission Forms* and *Assent Forms*. Please read and sign one of these forms. The **entire copies** of the *Permission* and *Assent Forms* must be returned to us in order for us to review your information. The second *Permission* and *Assent Forms* are for your files.

2. Please complete the *Patient Information Form* and return it to us.

3. Please complete the *Request for Medical Information Form* and return it to us. This form gives us your permission to communicate directly with your **child's** neurologist and/or primary care physician. We would be happy to request your medical records on your behalf once we receive this signed form.

4. Optional: If you would like to communicate with us via email, you must sign an additional consent form, the *Patient Email Consent Form*. This form is optional, but must be completed if you want to use email to communicate with National Registry staff. There are two copies of this form – return one signed copy to us. The second copy is for your files.

For your convenience, we have enclosed a postage paid envelope. Please place all signed forms into this envelope and return the packet to us. Once all of your information is received, it will be reviewed carefully. You may receive a phone call or letter from one of the study coordinators to clarify information.

If you have any questions as you complete this process, please do not hesitate to call us. Our toll free number is 1-888-925-4302. Once we have your signed email consent form, you may email us at dystrophy_registry@URMC.rochester.edu.

We sincerely appreciate your willingness to participate in this important endeavor!

Sincerely,

James Hilbert, MS
Health Project Coordinator

Elizabeth Luebbe
Health Project Coordinator

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

ASSENT FORM
(**Adolescents ages 13-17 years)**

Principal Investigator: Rabi Tawil, M.D. Study
Coordinator: James Hilbert, M.S.
Study Coordinator: Elizabeth Luebbe, M.S.

What are some things you should know about research studies?
You are being asked to take part in a study. Your parent or guardian needs to give permission for you to be in this study. You do not have to be in this study if you don't want to, even if your parent has given permission. You can choose whether or not to be in this study. You may
decide not to join. Or, if you join, you may decide to stop being in the study, at any time, for any reason,.

What is the purpose of this study?
Research is how we often learn new things. The purpose of this study is to join a Registry that may help doctors and scientists learn about ways to help people with two muscle diseases. The two muscle diseases are myotonic dystrophy and facioscapulohumeral muscular dystrophy. A registry is a place where medical information is collected and studied for medical research.

You are being asked to join because you or somebody in your family has one of these muscle diseases. The goals of the Registry are to:

- To keep track of people with muscle problems.
- To share information with doctors and scientists so that they can learn more about the cause of muscle problems and develop better treatments. We won't share your name or any information that could identify you.
- To help doctors and scientists find people with muscle problems to participate in their studies. You and your parents can choose whether or not to join any other studies. You don't have to join any other studies.
- To learn more about families with muscle problems.

What will happen if you take part in the study?
If you decide to take part in this study, you will be asked to help your parents answer questions about your symptoms or problems. People without these muscle diseases will answer a few questions about muscle diseases in their family. We will collect information from your doctor to learn more about your symptoms if you have a muscle disease. We will also collect information from your doctor if you had test that says you don't have a muscle disease.

For office use only: Registry Number: _____
RSRB Approval Date: 9/11/2020
Expiration Date: 9/10/2021

If you decide to join the Registry, you may be asked at a later time if you would like to help with other studies about these muscle diseases. We will send a letter through the mail, email, or online to describe these studies. You can review the information with your parents and decide
if you want to help with these studies too. No other doctor or research will know you are in the Registry. It will be up to you and your parents to talk to the other doctors or researchers. We keep your name private and let you decide about what other studies to join.

We will also send you a newsletter through the mail, email, or online with new information about research and muscle diseases.

How long will you be in this study?
Your participation in this study may last for several years. We will send you a new questionnaire each year to see if you have any changes (new address, new phone number, or new symptoms if you have a muscle disease). These forms help us keep track of how muscle diseases change over time.

Who will be told the things we learn about you in this study?
The information we collect about you will be kept private. Some of your information may be shared with other researchers, but this information won't include your name or anything that could identify you.

What are the possible risks or discomforts involved from being in this study?
The Registry includes questions that you may feel uncomfortable. You do not have to share any information you do not want to. There may also be an accidental release of your information to other groups. We have many rules to help prevent such accidents.

The University of Rochester makes every effort to keep the information collected from you private. In order to do so, we follow governmental laws about privacy, lock our computers and files, and have other safety tools. Sometimes, however, researchers need to share information that may identify you with people that work for the University, the government or the study sponsor. If this does happen we will take precautions to protect the information you have provided. Results of the research may be presented at meetings or in publications, but your name will not be used.

What are the possible benefits from being in this study?
The potential benefit to you from being in the Registry is receiving information about studies you may want to join. You will also receive newsletters and other information about muscle diseases.

What if you or your parents don't want to be in this study?
You do not have to sign this form if you don't want to be in the Registry. Even if your parents say yes, you do not have to. You can change your mind at any time. If someday you decide you want your name taken off the Registry list, just tell your parents or call us and we will remove your name. No one will be upset with you.

Will you get any money or gifts for being in this study?
You will not be paid or given anything for being in this study.

What if you have questions about this study?
For more information concerning this research or if you feel that being in the study has resulted in any research related injury, emotional or physical discomfort, please contact:

James Hilbert, MS or Elizabeth Luebbe, MS
University of Rochester, Department of
Neurology
601 Elmwood Ave, Box 673
Rochester, NY 14642
Telephone: (888) 925-4302 or (585) 276-0004.

What if you have questions about your rights as a research subject?
Please contact the University of Rochester Research Subjects Review Board at 265 Crittenden Blvd., CU 420315, Suite 1-250, Rochester, NY 14642-8315, Telephone (585) 273-4127 or (877) 449-4441 for the following reasons:

- You wish to talk to someone other than the research staff about your rights as a research subject;
- To voice concerns about the research;
- To provide input concerning the research process;
- In the event the study staff could not be reached.

Do I have to be in this study?
Taking part in this study is your choice. You are free not to take part or to stop at any time, for whatever reason. No matter what decision you make, there will be no penalty to you. In the event that you do stop this study, the information you have already provided will be kept private.

SIGNATURE/DATES

SUBJECT ASSENT
I have received two identical copies of this form (one to keep and one to return). I have read this form. If I had any questions, I have called the study team and have received the answers to my questions. I agree to participate in this study.

CHILD'S PRINTED NAME: _____

CHILD'S SIGNATURE: _____

DATE: _____

PERSON OBTAINING CONSENT
The subject has been given adequate opportunity to read the consent before signing and has been provided with a copy of the consent form for his/her records.

REGISTRY COORDINATOR PRINTED NAME: _____

REGISTRY COORDINATOR'S SIGNATURE: _____

DATE: _____

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

Permission Form
(Must be signed by a parent or guardian)

Principal Investigator: Rabi Tawil, M.D.
Study Coordinator: James Hilbert, MS
Study Coordinator: Elizabeth Luebbe, MS

This consent form describes a study, what you may expect if you decide to have your child take part, and important information to help you make your decision. Please read this form carefully. Please ask questions about anything that is not clear before you agree to participate.

Please note:
- Being in this study is voluntary – it is your choice.
- If your child joins this study, you can change your mind and stop at any time.
- There are risks from participating and you should understand what these mean to you.

INTRODUCTION

Your child is being asked to participate in this study because he/she or a family member has myotonic dystrophy (DM), facioscapulohumeral muscular dystrophy (FSHD), or a related disease (whose symptoms are similar to those of DM or FSHD).

This study has been established at the University of Rochester with the support from the National Institutes of Health (NIH). The study has been operational since September 2000 and is being conducted by Dr. Tawil of the University of Rochester's Department of Neurology (Rochester, NY).

PURPOSE

The purpose of this study is to collect information about the symptoms of DM and FSHD and to connect patients with researchers. This study's main goals are to:
1) Help researchers collect and study accurate, firsthand information on how DM and FSHD affect people;
2) Help researchers recruit patients with DM and FSHD into clinical trials;
3) Share information about exciting opportunities and advances in DM and FSHD with patients, care providers, and researchers.

What do I do?

1.) Complete questionnaires and other forms on DM and FSHD

Participation involves completing questionnaires about DM and FSHD in your child. If your child is unaffected, you will complete a shortened questionnaire. With your permission, we will

request your child's medical records about their DM or FSHD. Once your child is enrolled, the study staff will send you a questionnaire through mail, email, or social media each year to update his or her information.

One way the study helps advance knowledge of DM and FSHD is by sharing this detailed medical and other information about patients with other researchers, ***while still protecting your child's privacy***. This is done by hiding any information that could identify your child (name, address, etc.) from the researchers. The information that we share with researchers is "de-identified" because all personal identifiers have been removed. Your child's personal information such as their name, address, or other information that identifies him/her or your family will be labeled with a code number, stored in a secure place, and protected with a password. Only authorized people who work on the study will have access to this code and protected information.
Your child's identifiable information will not be shared with anyone outside the Registry (unless you give your permission to share it). Approved researchers will be allowed to see and study only de-identified information (information that has been removed of all identifiers). They can analyze this de-identified information to study the most common symptoms in DM and FSHD, learn how they progress over time, and other topics to better understand these rare diseases and to develop new treatments.

A subset of de-identified information collected from your child may be shared with certain other databases. We may share de-identified information with other registries that collect information on many rare disease and registries specific for DM and FSHD in Germany, Italy, and Australia for example. We may share de-identified information with other databases in order to develop global knowledge of DM and FSHD that may lead to new research studies, clinical trials, and clinical treatments.

2.) Receive information about other studies and decide if you want to join
Another way the Registry supports research is by helping recruit patients who may be eligible to participate in clinical studies. These studies may involve traveling to research centers across the US or completing questionnaires at home. Unaffected family members may be recruited to complete studies on quality of life or to serve as "healthy controls" to compare muscles strength or other things to people with DM or FSHD.

Approved researchers can recruit members of the Registry who look like a good match for their study. If your child is a good match, you will be provided with a description of the study through mail, email, or social media and given information on how to contact the researchers (their name, phone number, etc.). It is up to you to choose whether or not to contact the researchers. The researcher cannot contact you directly.

3.) Receive newsletters and other updates
We provide newsletters and other updates through mail, email, or social media with exciting news in DM and FSHD research and clinical care. You can specify how you'd like to receive this information.

Who is conducting this study?
Dr. Rabi Tawil is the Principal Investigator. Drs. Mike McDermott and Charles Thornton are Co-Investigators.

This study is also guided by members of its Scientific Advisory Committee. Members of this committee include scientists and researchers from all over the United States and Canada, who were instrumental to the development and design of the forms of the Registry and who provide ongoing support to review research applications submitted to the Registry. Researchers all over the world can use the Registry.

Is your child's information safe and confidential?
Yes! We follow stringent federal, state, and University guidelines to project privacy. Examples include the federal Health Insurance Portability and Accountability Act (HIPAA) which provides guidelines for maintaining privacy and the security of health information. All requests from other researchers or databases to use information from the Registry are reviewed by our Scientific Advisory Committee. Other safeguards are discussed on pages 5 and 6.

How do researchers apply to the Registry?
Researchers interested in using the Registry to analyze information or recruit patients submit a brief application and summary of their study. These studies are reviewed by the Registry's Scientific Advisory Committee. Studies are reviewed for safety, privacy, and scientific purpose. Upon approval of a research study, the Registry staff will work with the researcher to determine which members to recruit based on the number of subjects needed, the inclusion and exclusion criteria detailed in the study, geographical restrictions, etc. Eligible members will be sent an announcement through mail, email, or social media regarding the approved study and can volunteer to participate by contacting the researcher.

What types of studies are available?
Some studies involve filling out questionnaires at home, for example, about quality of life and other important topics. Other studies may involve collecting blood or tissue samples, testing your child's muscles, or testing new treatments. Each study is voluntary and requires your permission (consent) for your child to participate.

REGISTRY PROCEDURES

The forms you have received will take about 20 minutes to read and complete. The following information is requested:

1. This **entire** permission form (pages 1-7) with a signature from you or your child's legal guardian. In addition, an assent form will be read and signed by children aged 13 to 17 who are able to understand the procedures involved in the Registry.
2. A Patient Information Form with your child's name, address and phone number, as well as information about your child's muscle strength, general health, and how your child's disease has affected his or her daily life. A shortened version of this form will be completed for children who are unaffected.
3. A Request for Medical Information form. Please provide the complete name, address, and phone number of your child's doctors on this form. This form gives us permission to request medical records about your child's disease and how it was diagnosed. This form permits your child's physician to send test results such as the results of muscle biopsy, genetic testing, heart tracing, electromyography (EMG), as well as records that pertain to your child's muscular dystrophy. We will only request this information from

unaffected children if they have received a genetic test or other exams that show that they don't have muscular dystrophy.

4. If you would like to communicate with us via email you must sign an additional consent form, the Patient Email Consent Form.

- Please mail all completed forms to us in the self-addressed and pre-stamped envelope.
- Once we receive your child's application, we will review the forms and may contact you if additional information is needed.

After joining the Registry

- Once your child is enrolled into the Registry, we may contact you through mail, email, or social media about opportunities for your child to participate in studies. We will send you information about the studies for which your child may be eligible, including the researchers' names and phone numbers.

- If you are interested, you can contact the researcher for more information about the study. **The Registry will not provide information that could identify you or your child to the researcher.** All studies will have been reviewed and approved by the researcher's human subjects institutional review board and the Registry Scientific Advisory Committee.

- Once a year, Registry staff will send you a form through the mail, email, or social media to update changes to your/your child's address, phone number, and information about your child's health and symptoms of their muscle disease. The staff will also ask you about any studies in which your child has participated. It should take about 15 minutes to review and complete this "annual update" form.

- We ask that you contact us if your child move or if there is a change in his or her contact information so that we are able to update their file.

- Participation of family members is strongly encouraged. However, while family relationships may be recorded in the Registry none of their names or identifying information will be collected. No information about your child will be shared with members of your family. Each interested family member is encouraged to enter the Registry and to complete the forms themselves if interested and able.

NUMBER OF SUBJECTS

We expect 3,000 subjects or more to participate in this study.

BENEFITS OF PARTICIPATION

Your child might not benefit from being in this study. The potential benefit to your child from being in the Registry is receiving information about other studies that he or she may want to join. Your child will receive information about Registry activities and research advances in myotonic dystrophy, FSHD, and related diseases.

RISKS OF PARTICIPATION

There is minimal risk in taking part in this study. It includes questions that can be sensitive and that you or child may feel uncomfortable answering. You do not have to share any information

you do not want to. Another risk of participation is the possible loss of confidentiality due to unauthorized release of medical information.

SPONSOR SUPPORT

The University of Rochester is receiving payment from the National Institutes of Health (NIH) for conducting this study (grant #U54-NS048843 and contracts #N01-AR-5-2274 and #NO1-AR-0-2250).

COSTS

There will be no cost to you or your child to participate in this study.

PAYMENTS

You or your child will not be paid for participating in this study.

CERTIFICATE OF CONFIDENTIALITY

To help us protect your child's privacy, we have obtained a Certificate of Confidentiality from the Department of Health and Human Services (DHHS). With this Certificate, the investigators cannot be forced (for example, by court subpoena) to disclose research information that may identify your child in any Federal, State, or local civil, criminal, administrative, legislative, or other proceedings. Disclosure will be necessary, however, upon request of DHHS for audit or program evaluation purposes.

You should understand that a Certificate of Confidentiality does not prevent you or a member of your family from voluntarily releasing information about yourself or your involvement in this research. If an insurer, employer, or other person obtains your written consent to receive research information, then the investigator may not use the Certificate of Confidentiality to withhold this information. This means that you and your family must also actively protect your own privacy.

Finally, you should understand that the investigator is not prevented from taking steps, including reporting to authorities, to prevent serious harm to yourself or others.

CONFIDENTIALITY OF RECORDS AND HIPPA AUTHORIZATION

The University of Rochester makes every effort to keep the information collected from you private. In order to do so, we follow stringent federal, state, and University guidelines discussed above and below. For example, the University has sophisticated computer safeguards, such as firewalls, virus checking, network/workstation access passwords, and backup and disaster recovery. Paper forms are stored by unique Registry identification numbers, double locked, and maintained by other University safeguards.

While we will make every effort to keep information we learn about you private, this cannot be guaranteed. Other people may need to see the information. While they normally protect the privacy of the information, they may not be required to do so by law. Results of the research may be presented at meetings or in publications, but your name will not be used.

The federal Health Insurance Portability and Accountability Act (HIPAA) requires us to get your permission to use health information about you that we either create or use as part of the

research. This permission is called an Authorization. We will use related information from your medical records, results of laboratory tests, and both clinical and research observations made while you take part in the research.

We will use your health information to conduct the study, to determine research results, and possibly to develop new tests, procedures, and commercial products. Health information is used to report results of research to sponsors and federal regulators. It may be audited to make sure we are following regulations, policies and study plans. Strong Health policies let you see and copy this information after the study ends, but not until the study is completed. If you have never received a copy of the Strong Health HIPAA Notice, please ask the investigator for one.

To meet regulations or for reasons related to this research, the study investigator may share a copy of this consent form and records that identify you with the following people: The Department of Health and Human Services; the University of Rochester; and The National Institutes of Health.

If you decide to take part, your Authorization for this study will not expire unless you cancel (revoke) it. The information collected during your participation will be kept indefinitely. You can always cancel this Authorization by writing to the study investigator. If you cancel your Authorization, you will also be removed from the study. However, standard medical care and any other benefits to which you are otherwise entitled will not be affected. Canceling your Authorization only affects uses and sharing of information after the study investigator gets your written request. Information gathered before then may need to be used and given to others. For example, by Federal law, we must send study information to the FDA for drug and device studies it regulates. Information that may need to be reported to FDA cannot be removed from your research records.

As stated in the section on Voluntary Participation in the Consent Form, you can also refuse to sign this consent/Authorization and not be part of the study. You can also tell us you want to leave the study at any time without canceling the Authorization. By signing this consent form, you give us permission to use and/or share your health information as stated above.

The information for this Registry is collected under the authority of Sections 435-442 of the PHS Act (285d-285d-7 of Title 42, USC). The data will be maintained in accordance with the Privacy Act 42 United States Code 241.

CONTACT PERSONS

For more information about this research study please contact:

James Hilbert, MS or Elizabeth Luebbe, MS
University of Rochester, Department of Neurology
601 Elmwood Ave, Box 673
Rochester, NY 14642
Telephone: (888) 925-4302 or (585) 506-0004.

Please contact the University of Rochester Research Subjects Review Board at 265 Crittenden Blvd., CU 420315, Suite 1-250, Rochester, NY 14642-8315, Telephone (585) 273-4127 or (877) 449-4441 for the following reasons:

- You wish to talk to someone other than the research staff about your rights as a research subject;
- To voice concerns about the research;
- To provide input concerning the research process;
- In the event the study staff could not be reached.

VOLUNTARY PARTICIPATION

Taking part in this study is voluntary. Your child is free not to take part or to withdraw at any time, for whatever reason. No matter what decision you make, there will be no penalty or loss of benefit to which your child is entitled. In the event that your child does withdraw from this study, the information you have already provided will be kept in a confidential manner.

SIGNATURE/DATES

After reading and discussing the information in this consent form you should understand:

- Why this study is being done;
- What will happen during the study;
- Any possible risks and benefits to you;
- How your child's personal information will be protected;
- What to do if you have problems or questions about this study.

PARENT OR GUARDIAN PERMISSION

I have received two identical copies of this consent form (one to keep and one to return) and have read the contents. If I had any questions, I have called the study team and have received the answers to my questions. I agree to participate in this study. **After signing one copy** of this consent form, I will **mail the entire form** to:

Health Project Coordinator
National Registry of DM and FSHD
University of Rochester, Department of Neurology
601 Elmwood Ave, Box 673
Rochester, NY 14642

CHILD'S PRINTED NAME: _____

PARENT/GUARDIAN'S PRINTED NAME: _____

PARENT/GUARDIAN'S SIGNATURE: _____

DATE:_____

PERSON OBTAINING CONSENT

The subject has been given adequate opportunity to read the consent before signing and has been provided with a copy of the consent form for their records.

REGISTRY COORDINATOR PRINTED NAME: _____

REGISTRY COORDINATOR'S SIGNATURE: _____

DATE: _____

University of Rochester Department of Neurology
National Registry for DM and FSHD
601 Elmwood Ave, Box 673
Rochester, NY 14642
Telephone: (888) 925-4302 Fax: (585-276-2494)

UNIVERSITY of ROCHESTER MEDICAL CENTER

& Affiliates

SH 48 Authorization for Release/Disclosure of Medical and/or Behavioral Health Information

PLEASE PRINT.

Patient name: _ Date of Birth: _ _ _ _ _ _ _ _ _ _ _

Address: Patient's phone#:

City/State/Zip:

This Authorization allows URMC & Affiliates to: (check one or both)

☐ **SEND** copies of your record to (or discuss your information with) the provider/person/facility bel°""

☑ **RECEIVE** copies of your record from (or discuss your information with) the provider/person/facility bel°""

Name of Provider/ Person/Facility

Address

City, State, Zip Code

Phone #/Fax# include area code

PURPOSE FOR THIS REQUEST: ☐ Healthcare or Appointment **(date)**___ _ ☐ Insurance ☑ Other

TYPE OF RECORDS or INFORMATION REQUESTED: Check all that apply:

The records requested are to include: ☐ Mental Health Treatment Records ☐ Alcohol/Drug Treatment Records

(Release/disclosure of HIV-related information requires additional authorization on form NYS DOH2557 or OCA 960)

☐ **Inpatient admission(s)/date(s):**
(Check only one of the following 3 choices if requesting inpatient records)

☐ Treatment summary (includes discharge summary, history/physical, laboratory tests, x-ray reports, operative reports, pathology)

☐ Specific information or reports (describe): _____

☐ Other (describe): _____

☑ Outpatient/Office visits--date(s): All and/or specific illness/injury: muscle disease
(Check type of outpatient visit to be released)

☑ Clinic/doctor/dental visit ☐ Ambulatory Surgery visit ☐ Emergency Department Record
☐ Radiology report(s) ☑ Laboratory test results ☐ Immunizations ☑ Physical/occupational therapy record(s)
☑ Other (describe): Recent H&P, DNA results related to MD

AUTHORIZATION VALID FOR: (If nothing is checked below, this authorization is valid for this request only.)

☐ This request only

☑ One year from the date of this authorization **OR** _____(insert date) This authorization applies to the records of the treatment received on or prior to the date of this authorization.

☐ This request **and** for medical records of any **future** treatment of the type described above until: (insert date)

I understand that:

- My right to healthcare treatment is not conditioned on this authorization, except in very limited circumstances (e.g. non-emergent mental health or chemical dependency treatment).
- I may cancel this authorization at any time by submitting a *written* request to the address provided at the top of this form, except where a disclosure has already been made in reliance on my prior authorization.
- If the person or facility receiving this information is not a health care or medical insurance provider covered by privacy regulations, the information stated above could be redisclosed, except that chemical dependency treatment records protected by Federal Confidentiality Rules 42C R Part 2 may not be disclosed without my written authorization unless otherwise provided for in the regulations.
- There may be a charge for the requested records.
- The medical records requested above may be faxed in cases of medical necessity.

Signature of Patient or Representative _____ Date _____

Relationship to Patient (if Representative) ----------

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

Patient Information Form ***for individuals with Myotonic Dystrophy or Related Diseases***

The purpose of this form is to collect information from individuals who have myotonic dystrophy or a related disease. **Please return this form within three weeks if at all possible.** If you have any questions about this form, please call Local: (585) 506-0004, in Rochester NY or Toll Free: (888) 925-4302 for assistance.

If you have a disease that is related to myotonic dystrophy, such as proximal myotonic myopathy (PROMM) or myotonic dystrophy type 2, please write your diagnosis here _____ and then fill out the form even though all questions may not apply to your condition.

Date: _____

NAME:_____
First Middle (Maiden) Last

ADDRESS:_____
Street

_____
City State Zip Code

TELEPHONE: Home: (____) _____ Work: (____)_____
Area Code Number Area Code Number

EMAIL ADDRESS: _____

Date of Birth:____/____/_____ Sex: ☐ Male ☐ Female
Mo Day Year

Where did you learn about the Registry?
☐ Your doctor ☐ Internet ☐ MDA
☐ Family ☐ Support group ☐ Magazine/Newsletter
☐ Friend
☐ Other_____

For office use only. Name: _____ Registry Number: _____

INFORMATION ABOUT YOUR DIAGNOSIS OF MYOTONIC DYSTROPHY:

1. What was the first symptom of myotonic dystrophy?_____
_____

2. How old were you when you had your first symptom of myotonic dystrophy? (Give your best estimate even if you are not sure.)_____ years old.

3. How old were you when your myotonic dystrophy was diagnosed? (Give your best estimate even if you are not sure.)_____ years old.

4. Did you have any of these tests?

Examination by a neurologist	□ Yes	□ No	□ Not sure
Electromyography (EMG, needle inserted into muscles to check electrical activity)	□ Yes	□ No	□ Not sure
Muscle biopsy	□ Yes	□ No	□ Not sure
DNA test (blood test) for myotonic dystrophy	□ Yes	□ No	□ Not sure

5. Who made your diagnosis of myotonic dystrophy? (Check as many as apply)
   - □ primary care physician
   - □ a neurologist
   - □ family member
   - □ yourself
   - □ a specialist in a neuromuscular clinic or Muscular Dystrophy Clinic

6. Were you the first person in your family to have the diagnosis of myotonic dystrophy?
   □ Yes □ No □ Not sure

7.	**YES**	**NO**	**Not Sure**	
Is anyone else in your family affected with myotonic dystrophy? **If yes,** please indicate with a check in the appropriate boxes below.				
	YES	**NO**	**Not Sure**	**Number Affected**
Brothers and sisters				
Children				
(If yes, are any affected children under the age of 18? □ yes □ no)				
Mother				
Father				
Grandparents				
Aunts or uncles				
Cousins or other relatives				

8. Are any other members of your family in the Registry?
   □ Yes □ No □ Not sure

OCCUPATION AND EMPLOYMENT

What is your current occupation (complete below)

- ☐ Employed (describe your job)_____
- ☐ Homemaker ☐ Student ☐ Retired
- ☐ Disabled because of myotonic dystrophy ☐ Disabled (not due to myotonic dystrophy)
- ☐ Unemployed (not due to disability)

Comments_____

Has myotonic dystrophy affected your employment? ☐ Yes ☐ No

If yes, how (check boxes)

- ☐ Lost job
- ☐ Job modified to accommodate your physical limitations
- ☐ Forced to go on disability
- ☐ Early retirement

EDUCATION

Highest level of education completed: (check appropriate box)

- ☐ No formal education
- ☐ Grade school
- ☐ High school
- ☐ Technical school
- ☐ College
- ☐ Graduate school
- ☐ Other_____
- ☐ Don't know

USE OF ASSISTIVE DEVICES				**Your age when you started using the device (give your best estimate even if you are not sure).**
	YES	NO		Years old
Use ankle braces				Years old
Use long leg braces				Years old
Use a cane at times				Years old
Use a walker at times				Years old
Use a wheelchair. **If yes,** circle one: 1. For long distances only 2. Usually 3. Always				Years old
Use of CPAP or BIPAP for breathing assistance				Years old
Use ventilator				Years old
Have a pacemaker				Years old
Other_____				Years old

SIGNS AND SYMPTOMS

Do you have any of the following?					**Your age when the problem began (give your best estimate even if you are not sure).**
	Yes	No	Not sure		
Trouble with your hands/grip locking up, or hand stiffness					
Difficulty making a tight fist, loss of grip strength or difficulty opening jars					
Trouble speaking clearly					
Trouble with swallowing					
Weakness of face					
Difficulty walking on your toes or heels, or ankle weakness					
Difficulty getting up from the floor, rising from a chair, or climbing stairs					
Trouble with breathing or shortness of breath					
Cataracts					
Racing heart beat, irregular heart beat, palpitations, or pacemaker					
Baldness					

BROKEN BONES AND SURGERY

Have you ever had a broken bone or operation? ☐ Yes ☐ No
If yes, please list them and the date they occurred. *If you need more room, please use the back of this form.*

Broken bone or operation	**Year that it occurred**

MEDICATIONS

Do you take medications? □ Yes □ No □ Don't know
If yes, please give the name of each medication. Include both prescription and non-prescription drugs and herbal remedies.

Codes: 1 Have taken for less than one month
2 Have taken for one month to one year
3 Have taken for more than one year

Name of medication	Circle one			Daily Dosage Milligrams/Tablet	Daily Dosage Tablets/Day
	1	2	3		
	1	2	3		
	1	2	3		
	1	2	3		
	1	2	3		
	1	2	3		
	1	2	3		
	1	2	3		
	1	2	3		
	1	2	3		

If you need more room, please use the back of this form.

What is your current height:_____feet_____inches, and weight:_____pounds

ALLERGIES

Please list any foods or drugs to which you are allergic:

_____ _____
_____ _____
_____ _____
_____ _____

Do you smoke tobacco? □ Yes □ No

TREATMENTS OR COUNSELING

Have you ever received any of the following?

	Yes	No	Not sure
Physical therapy			
Genetic counseling			
Emotional or psychological counseling			
Speech therapy			
Occupational therapy			
Vocational rehabilitation			
Other_____			

OTHER MEDICAL PROBLEMS

Have you ever had or do you have any of these conditions:

- ☐ Diabetes
- ☐ High blood pressure
- ☐ Asthma
- ☐ Rheumatoid arthritis
- ☐ Emphysema
- ☐ Pneumonia
- ☐ Heart disease or heart beat irregularity
- ☐ Cancer or tumor, type_____
- ☐ High cholesterol
- ☐ Miscarriage
- ☐ Stillbirth
- ☐ Child showing signs of myotonic dystrophy within the 1st four weeks of life
- ☐ Psychological problems such as depression or anxiety
- ☐ Other_____
- ☐ Stroke
- ☐ Kidney trouble
- ☐ Thyroid trouble
- ☐ Stomach ulcers
- ☐ Gall bladder trouble
- ☐ Prostate trouble
- ☐ Liver trouble
- ☐ Chronic infection
- ☐ Trouble with sexual function
- ☐ Acid reflux or "heartburn"
- ☐ Constipation

ETHNICITY/RACE

Are you Hispanic or Latino? ☐ Yes ☐ No

How would you describe your race? Select one or more of the following categories:

- ☐ American Indian or Alaskan Native
- ☐ Black or African American
- ☐ Native Hawaiian or other Pacific Islander
- ☐ Asian
- ☐ White

SLEEP PROBLEMS

How likely are you to doze off or fall asleep in the following situations, in contrast to feeling just tired? This refers to your usual way of life in recent times. Even if you have not done some of these things recently try to work out how they would have affected you. Use the following scale to choose the *most appropriate number* for each situation:

0 = would *never* doze
1 = s*light* chance of dozing
2 = *moderate* chance of dozing
3 = *high* chance of dozing

Situation	Chance of dozing
Sitting and reading	
Watching TV	
Sitting, inactive in a public place (such as a theater or a meeting)	
As a passenger in a car for an hour without a break	
Lying down to rest in the afternoon when circumstances permit	
Sitting and talking to someone	
Sitting quietly after lunch without alcohol	
In a car, while stopped a few minutes in traffic	

Have you ever participated in a research study for myotonic dystrophy ?

□ Yes □ No □ Not sure

Have you ever received an experimental treatment for myotonic dystrophy?

□ Yes □ No

If yes, what was that treatment:_____

In case you needed help filling out this form, who was your helper? (state below)
Name of individual filling out the form:_____
Relationship to applicant:_____

Please provide the name, address, and telephone number of a family member or friend we can contact in case you move or change your phone number.

NAME:_____RELATIONSHIP:_____
ADDRESS:_____
PHONE NUMBER:_____

Medical records, which confirm your diagnosis, must be sent to us for review. Attached is a Request for Information form. If you sign it and return it to us, we can contact your doctor for any test results and they can send them directly to us.

IMPORTANT

Please read, sign and return the attached Consent Form. Without it we cannot consider you for entry into the Registry.

Thank you for your help with the Registry.

Local: (585) 506-0004, Rochester NY
Toll Free: (888) 925-4302
FAX: (585) 273-1255
Address: 601 Elmwood Avenue, Box 673, Rochester, NY 14642-8673

The information for this Registry is collected under the authority of Sections 435-442 of the PHS Act (285d-285d-7 of Title 42, USC). The data will be maintained in accordance with the Privacy Act 42 United States Code 241.

This project has been funded in whole or in part by the National Institutes of Health (grant #U54-NS048843 and contracts #N01-AR-5-2274 and #NO1-AR-0-2250).

UNIVERSITY of ROCHESTER MEDICAL CENTER

Research Subject E-mail Consent Form

Subject name:
Subject #:
Subject e-mail:
Researcher: Dr. Rabi Tawil
Researcher e-mail: dystrophy_registry@urmc.rochester.edu
Authorized representative:
Name: _____
Relationship: _____
E-Mail: _____

1. RISK OF USING E-MAIL

Transmitting subject information by e-mail has a number of risks that subjects should consider. These include, but are not limited to, the following:
a) E-mail can be circulated, forwarded, stored electronically and on paper, and broadcast to unintended recipients.
b) E-mail senders can easily misaddress an e-mail.
c) Backup copies of e-mail may exist even after the sender or the recipient has deleted his or her copy.
d) Employers and on-line services have a right to inspect e-mail transmitted through their systems.
e) E-mail can be intercepted, altered, forwarded, or used without authorization or detection.
f) E-mail can be used to introduce viruses into computer systems.

2. CONDITIONS FOR THE USE OF E-MAIL

The researcher cannot guarantee but will use reasonable means to maintain security and confidentiality of e-mail information sent and received. The subject and researcher must consent to the following conditions:
a) E-mail is not appropriate for urgent or emergency situations. The researcher cannot guarantee that any particular e-mail will be read and responded to.
b) E-mail must be concise. The subject should schedule an appointment if the issue is too complex or sensitive to discuss via e-mail.
c) E-mail communications between subject and researcher will be filed in the subject's research record.
d) The subject's messages may also be delegated to any member of the study team for response.
e) The researcher will not forward subject-identifiable e-mails outside of URMC and Affiliates without the subject's prior written consent, except as authorized or required by law.
f) The subject should not use e-mail for communication regarding sensitive medical information.
g) It is the subject's responsibility to follow up and/or schedule an appointment if warranted.
h) Recommended uses of subject-to-researcher e-mail should be limited to:
   a. Appointment requests
   b. Prescription refills
   c. Requests for information
   d. Updates to information or exchange of non-critical information such as laboratory values.

3. INSTRUCTIONS

To communicate by e-mail, the subject shall:
a) Avoid use of his/her employer's computer.
b) Put the subject's name in the body of the e-mail.
c) Put the topic (e.g., study question) in the subject line.
d) Inform the researcher of changes in the subject's e-mail address.
e) Take precautions to preserve the confidentiality of e-mail.
f) Contact the researcher's office via conventional communication methods (phone, fax, etc.) if the subject does not receive a reply within a reasonable period of time.

4. SUBJECT ACKNOWLEDGMENT AND AGREEMENT

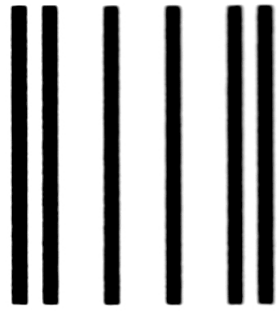
I acknowledge that I have read and fully understand this consent form. I understand the risks associated with the communication of e-mail between the researcher and me. I consent to the conditions and instructions outlined here, as well as any other instructions that the researcher may impose to communicate with me by e-mail. I agree to use only the pre-designated e-mail address specified above. Any questions I may have had were answered.

_____
Subject or Authorized Representative signature

Date

_____
Researcher signature

Date



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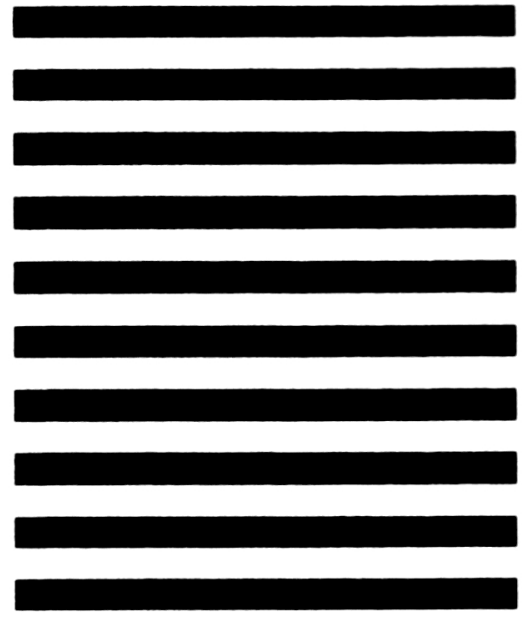

University of Rochester
University of Rochester Medical Center
National Registry, Box 673
PO Box 23029
Rochester NY 14692-6971