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**! The Registry connects people with myotonic dystrophy and FSHD with research opportunities. Anyone with myotonic dystrophy or FSHD is eligible to join, as well as family members.

Please complete the following enclosed forms to join the Registry:

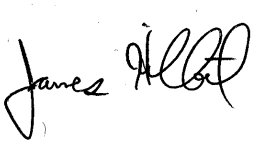
1. Consent Form - Please sign and return one copy. The second copy is for you to keep.
2. Assent Form – Completed if the enrollee is a child between the ages of 13-17 years old.
3. Patient Information Form

If you previously had a genetic test for myotonic dystrophy or FSHD, it would greatly help us to have a copy of the results. If you do not have a copy, you can request one from your doctor or the office that ordered the test. If possible, please send us a copy of your test results along with the forms above. This information will be added to your Registry record.

Please return the completed forms to us in the enclosed prepaid envelope. If you have any questions, please contact us at 1-888-925-4302 or at dystrophy_registry@URMC.rochester.edu.

We appreciate your support of research for DM and FSHD!

Sincerely,



James Hilbert, MS
Health Project Coordinator

Elizabeth Luebbe
Health Project Coordinator

Address: 601 Elmwood Avenue, Box 673, Rochester, NY 14642-8673
Phone: Toll-free 1-888-925-4302 Fax: 585-273-1255
Email: Dystrophy_registry@urmc.rochester.edu Website: www.dystrophyregistry.org
Facebook: www.facebook.com/NationalRegistryofMyotonicDystrophyandFSHD

RSRB Approval Date: 9/30/2024

CONSENT FORM

Study title: National Registry of Myotonic Dystrophy and Facioscapulohumeral Muscular Dystrophy Patients and Family Members

Principal Investigator: Johanna Hamel, MD

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

A person who takes part in a research study is called a research subject, or research participant. In this consent form, "you" generally refers to the research subject. If you are a parent/legal guardian for the potential subject, "you" in the rest of this form generally means your child or the adult who will be the research subject.

Key Information

- Being in this research study is voluntary – it is your choice.
- You are being asked to take part in this study because you or a family member has myotonic dystrophy (DM) or facioscapulohumeral muscular dystrophy (FSHD).
- The purpose of the National Registry is to collect information about the symptoms of DM and FSHD and to connect patients with researchers.
- Your participation in this study will last for the next 5-10 years or longer.
- Procedures include completing a questionnaire and providing updates to your information each year. You will also receive information about studies related to DM and FSHD and information on how to participate. You may also receive email and newsletters related to Registry activities.
- There are risks from participating.
  - The most common risk is that you may feel uncomfortable answering certain questions about your symptoms. You do not have to share any information that you do not want to.
  - One of the most serious risks is a possible loss of confidentiality due to the unauthorized release of medical information. See the "Risks of Participation" section in this consent form for more information. You should discuss these risks in detail with the study team if you have any questions.
- You might not benefit from being in this research study. A potential benefit is receiving information about studies that you may want to join and receiving updates on advances in DM and FSHD research and clinical care.

PURPOSE

The goals of this Registry are to:

- Help researchers collect and study information on how DM and FSHD affect people;
- Help researchers recruit patients with DM and FSHD into clinical studies and trials;
- Share information about opportunities and advances in DM and FSHD research with you, care providers, and researchers.

Registry Number: _____

RSRB Approval Date: 9/30/2024
Expiration Date: 8/27/2025

DESCRIPTION OF PROCEDURES

The forms for the Registry will take about 20 minutes to read and complete. You can complete the forms by paper or online through Research Electronic Data Capture (REDCap). REDCap is a secure, HIPAA-compliant, web-based application used for electronic capture and management of research and clinical study data. The following is requested to participate in the Registry:

- **Complete the "Patient Information Form" questionnaire.** This form will ask for your contact information as well as information about your muscle strength, general health, and how your muscular dystrophy affects your daily life. Unaffected family members will complete a shortened version of this form.

- **Optional procedures:**

  Optional to provide us with a copy of your genetic test results: If you previously had a genetic test for myotonic dystrophy or FSHD, it would help us to have a copy of the results. If you do not have a copy, you can request one from your doctor or the office that ordered the test. This information is important because sometimes researchers ask us to send notices only to people who have had a genetic test or whose testing showed a particular type of result. If possible, please send us a copy of your test results along with the forms above. This information will be added to your Registry record. You have the option to share a copy of your genetic test results, by indicating your consent at the end of this form.

  Optional to complete an Authorization for Release of Medical Information form. We may ask for your permission to obtain your medical records, for example, if there is not enough information to determine your diagnoses or clarify certain symptoms, like patterns of muscle weakness or non-muscle symptoms. If we request this information, you have the option to complete this Authorization form. If you are asked and agree to share, at that time, please provide the complete name, address, and phone number of one or two of your doctors on this form. This form gives us permission to request medical records about your muscular dystrophy and how it was diagnosed. This form permits your physician(s) to send test results such as the results of muscle biopsies, genetic testing, heart tracing (e.g., EKG), electromyography (EMG), as well as records that pertain to your muscular dystrophy. If you are an unaffected family member, we may only request this information if you have received a genetic test or other exams that show that you do not have muscular dystrophy. You have the option to sign this authorization, by indicating your consent at the end of this form.

- If you complete the forms on paper, please mail all completed forms to us in the enclosed, prepaid envelope. If you complete the forms online, you have the option to save and return later. When you click "save," you will receive an individualized Return Code to return and complete your application at a later time, if you choose.

- Once we receive your application through the mail or online, we will review your forms and may contact you if additional information is needed. You will receive a notification in the mail or email that all of your forms have been reviewed and that you are enrolled in the Registry.

After joining the Registry

- Once you are enrolled in the Registry, we may contact you through the mail or email about opportunities to participate in research studies. Some studies involve filling out questionnaires at

home about your quality of life. Other studies involve collecting blood or tissue samples, testing your muscle strength, or testing new treatments. Each study is voluntary and requires your agreement (consent).

- If you are interested in such studies, you can contact the researcher for more information about the study. **The Registry will not provide any information that could identify you to the researcher**. All research studies are reviewed and approved by the researcher's human subjects institutional review board and by the Scientific Advisory Committee of this Registry.

  Once a year, we will send you a form through the mail or email to update your address, phone number, and information about your health and/or any symptoms of your muscular dystrophy. It should take about 15 minutes to review and complete this form. Completion of the form is voluntary.

- We ask that you contact us if there are changes to your home address, phone number, or email address so that we are able to update your contact information.

- Participation of family members is strongly encouraged. No information about you will be shared with members of your family. Each family member is encouraged to enter the Registry and to complete the forms themselves, if interested and able.

- Scientists, researchers, and clinicians will be allowed to see and study Registry data that is de-identified or anonymous (information that cannot identify you). Researchers need to submit an application to the Registry team to get approval and receive data. They can analyze this de-identified information to study the symptoms in DM and FSHD, learn how symptoms progress over time, and other topics to better understand these diseases and to develop new treatments.

- A subset of de-identified information collected from you may be shared with certain other databases. We may share de-identified information with other national or international registries that collect information on multiple rare disease and registries that are specific to DM or FSHD. We may share de-identified information with other databases in order to increase global knowledge of DM and FSHD that may lead to new research studies, clinical trials, and clinical treatments. No information will be shared that could identify you.

NUMBER OF SUBJECTS

We expect 3,500 subjects or more to participate in this Registry.

BENEFITS OF PARTICIPATION

You might not benefit from being in this Registry. A potential benefit to you from being in the Registry is receiving information about other studies you may want to join. You will receive information about Registry activities and research advances in myotonic dystrophy, FSHD, and related diseases.

Researchers may benefit by using the Registry to study why individuals have different symptoms, learn about how certain treatments work, help medical professionals improve how they manage care for individuals with DM and FSHD, and advance research in DM and FSHD by analyzing de-identified Registry data.

RISKS OF PARTICIPATION
There is minimal risk in taking part in this Registry. Participation includes questions that can be sensitive and that may make you may feel uncomfortable. You do not have to share any information that you do not want to. Another risk of participation is the possible loss of confidentiality due to an unauthorized release of medical information.

SPONSOR SUPPORT
The University of Rochester is receiving payment from the National Institutes of Health (NIH) for conducting this research.

COSTS
There will be no cost to you to participate in this Registry.

PAYMENTS
You will not be paid for participating in this Registry.

CERTIFICATE OF CONFIDENTIALITY
To help us protect your privacy, we have a Certificate of Confidentiality from the National Institutes of Health (NIH). With this Certificate, the investigators cannot be forced (for example, by court subpoena) to disclose research information that may identify you 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 researcher is not prevented from taking steps, including reporting to authorities, to prevent serious harm to yourself or others.

Confidentiality of Records and Authorization to Use and Disclose Information for Research Purposes
The University of Rochester makes every effort to keep the information collected from you private. In order to do so, we have 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. Sometimes, however, researchers need to share information that may identify you with people that work for the University, regulators or the study sponsor.

If you have never received a copy of the University of Rochester Medical Center (URMC) and Affiliates Notice of Privacy Practices, please ask the investigator for one.

What information may be used and given to others?
The study doctor will get your personal and medical information. For example:

- Research records
- Records about phone calls made as part of this research

Who may use and give out information about you?

- The study doctor and the study staff
- URMC and Affiliates

Your information may be given to:

- The Department of Health and Human Services
- The University of Rochester
- The Registry's Scientific Advisory Committee, the National Institutes of Health, other government agencies, and foreign government regulatory agencies.

Why will this information be used and/or given to others?

- To do the research
- To study the results
- To see if the research was done correctly

If the results of this study are made public, information that identifies you will not be used.

What if I decide not to give permission to use and give out my health information?
Then you will not be able to be in this research study.
May I review or copy my information?
Yes, but only after the research is over.

How long will this permission be valid?
This permission will last indefinitely.

May I cancel my permission to use and disclose information?
Yes. You may cancel your permission to use and disclose your health information at any time. You do this by sending written notice to the study doctor. Upon receiving the written notice, the study team will no longer use or disclose your health information and you will not be able to stay in this study. Information that has already been gathered may need to be used and given to others for the validity of the study.

May I withdraw from the study?
Yes. If you withdraw your permission to be in the study, no new health information identifying you will be gathered after that date. Information that has already been gathered may still be used and given to others.

Is my health information protected after it has been given to others?
No. There is a risk that your information will be given to others without your permission.

Use of Email for Communication in Research

You have the option to receive communications about this study via email, by indicating your consent at the end of this form. Messages will be limited to clarification about the forms you completed, annual requests to update your information, general correspondence, announcements about opportunities to participate in other research, and newsletters or general information.

Email may be sent or received in an unencrypted (unprotected) manner. Therefore, there is a risk that the content of the communication, including your personal information, could be shared beyond you and the research team. Your consent below indicates that you understand this risk. The University of

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Rochester is not responsible for any interception of messages sent through email or texting. Email communications between you and the research team may be filed in your research record.

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
Email: dystrophy_registry@urmc.rochester.edu
Telephone: (888) 925-4302 or (585) 276-0004.

Please contact the University of Rochester Research Subjects Review Board at 265 Crittenden Blvd., CU 420628, Rochester, NY 14642, Telephone (585) 276-0005 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. You are 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 you are entitled. In the event that you do withdraw from this study, the information you have already provided will be kept in a confidential manner.

Optional Research Activities:

Place your initials in the YES **OR** NO box, based upon your decision to take part.

Communication with the Study Team

YES *(initial)*	**NO** *(initial)*	*I consent to the use of **email** in this study. If yes, enter email address:* _____

Share a copy of your genetic test result if available.

YES *(initial)*	**NO** *(initial)*

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Sign an authorization for release of medical information if asked by Registry staff

YES *(initial)*	**NO** *(initial)*

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 personal information will be protected;

What to do if you have problems or questions about this study.

Please complete section 1 **OR** section 2.

1.) SUBJECT CONSENT (For participants 18 years or older and capable of providing consent)

I have read (or it has been read to me) the contents of this consent form and have been encouraged to ask questions. If I had any questions, I have asked the study team and have received the answers to my questions. I agree to participate in this study.

If completing these forms on paper, I have received two copies of this consent form (one copy to return to the study team and the other copy for my records and future reference). If completing these forms online, I will receive an email with a copy of this form for my records and future reference.

_____
Subject Name (Printed by Subject)

_____ _____
Signature of Subject Date

2.) CONSENT FROM PARENT, LEGAL GUARDIAN, or LEGALLY AUTHORIZED REPRESENTATIVE (LAR)

I have read (or it has been read to me) the contents of this consent form and have been encouraged to ask questions. If I had any questions, I have asked the study team and have received the answers to my questions. I agree to allow the subject to participate in this study.

If completing these forms on paper, I have received two copies of this consent form (one copy to return to the study team and the other copy for my records and future reference). If completing these forms online, I will receive an email with a copy of this form for my records and future reference.

_____
Subject Name (Printed by parent, guardian, or LAR)

_____
Name of Parent, Guardian, or LAR (Printed)

_____ _____
Signature of Parent, Guardian, or LAR Date

Registry Number: _____

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Below Completed by Registry Staff Only

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:_____

Registry Number: _____

RSRB Approval Date: 9/30/2024
Expiration Date: 8/27/2025

ASSENT FORM
(Adolescents ages 13-17 years)

Study title: National Registry of Myotonic Dystrophy and Facioscapulohumeral Muscular Dystrophy Patients and Family Members

Principal Investigator: Johanna Hamel, M.D.

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. 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 problems. The two muscle problems are myotonic dystrophy and facioscapulohumeral muscular dystrophy (or FSHD). 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 problems. 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 problems will answer a few questions about their family. We may collect information from your doctor to learn more about your symptoms if you have a muscle problem. We may also collect information from your doctor if you had test that says you don't have a muscle problem. You have the option to share some of your medical record with us.

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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 problems. 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 or guardian 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 problems.

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 problem). These forms help us keep track of how muscle problems 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 may make you 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 steps to protect the information that 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 problems.

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 or guardian say yes, you do not have to. You can change your mind at any time. If some day you decide you want your name taken off the Registry list, just tell your parents or guardian 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:

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

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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 420628, Rochester, NY 14642, Telephone (585) 276-0005 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 read (or it has been read to me) the contents of this consent form and have been encouraged to ask questions. 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.

If completing these forms on paper, I have received two copies of this consent form (one to return to the study team and the other copy for my records and future reference). If completing these forms online, I will receive an email with a copy of this form for my records and future reference.

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 will be provided with a copy of the consent form for his/her records.

REGISTRY COORDINATOR PRINTED NAME: _____

REGISTRY COORDINATOR'S SIGNATURE: _____

DATE:_____

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

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Expiration Date: 8/27/2025

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

Patient Information Form: *Myotonic Dystrophy*

What is your diagnosis?

☐ Myotonic dystrophy type 1 (DM1) ☐ Myotonic dystrophy type 2 (DM2) ☐ Unknown

Date: _____

Name: _____
First Middle Last (Maiden)

Address: _____
Street

_____
City State Zip Code

Telephone: Home: ____ - _____ - _____ Cell: ____ - _____- _____

Work: ____- _____- _____

Email address: _____

Date of birth: ____ / ____ / _____
Month Day Year

Sex at birth: ☐ Male ☐ Female **Gender:** ☐ Male ☐ Female ☐ Other

Ethnicity: (Check one with which you most closely identify)

☐ Hispanic or Latino ☐ Not Hispanic or Latino

Race: (Check all that apply)

☐ American Indian or Alaskan Native ☐ Asian ☐ Black or African American

☐ Native Hawaiian or other Pacific Islander ☐ White

Current Height: ____ feet _____ inches **Current Weight:** _____ pounds

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For office use only. Registry # : _____ Entered by: _____ Verified by: _____

Where did you learn about this Registry?

☐ Your doctor ☐ Internet ☐ MDA
☐ Family ☐ Magazine/Newsletter ☐ Support group
☐ Friend ☐ Other _____

MYOTONIC DYSTROPHY ONSET AND DIAGNOSIS

What was your first symptom of myotonic dystrophy?_____

_____

How old were you when you had your first symptom of myotonic dystrophy? _____ years old
(Estimate if not sure)

How old were you when your myotonic dystrophy was diagnosed? _____ years old
(Estimate if not sure)

Have you had any of these tests?

Examination by a neurologist . . . . . ☐ Yes ☐ No ☐ Not sure

Electromyography . . . . . ☐ Yes ☐ No ☐ Not sure
(EMG: needle inserted into muscles to check electrical activity)

Muscle biopsy . . . . . ☐ Yes ☐ No ☐ Not sure

DNA test (blood test) for myotonic dystrophy ☐ Yes ☐ No ☐ Not sure

Who made your diagnosis of myotonic dystrophy? (Check as many as apply)

☐ Primary care physician ☐ Neurologist ☐ Yourself ☐ Family member
☐ Specialist in neuromuscular clinic or muscular dystrophy clinic

FAMILY HISTORY

Are you the **first** person in your family to have the diagnosis of myotonic dystrophy?

☐ Yes ☐ No ☐ Not sure

Are other members of your family in this Registry? ☐ Yes ☐ No ☐ Not sure

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In the table below, indicate any blood relatives, alive or deceased, that have or had myotonic dystrophy (DM).

If you are adopted, please check here ☐ and complete the information in the table below for your biological family (if known).

	Had/has DM	**Unaffected**	**Unsure**
	Check appropriate boxes below		
Mother			
Father			
	Number with DM	**Number without DM**	**Number Unsure**
Grandparents			
Children			
Grandchildren			
Siblings			
Half-Siblings			
Aunts or uncles			
Other (specify below):			

Have any of your children shown signs of myotonic dystrophy within the 1st four weeks of life (e.g., trouble breathing, trouble eating, and muscle weakness)?

☐ Yes ☐ No ☐ Not applicable

EDUCATION

School status:

☐ No formal education
☐ Going to school (indicate grade level - kindergarten through 12th grade): _____
☐ Going to college (indicate year or level): _____
☐ Not currently in school or college

Highest level of education completed: (Check appropriate box)

☐ Grade school
☐ High school, GED or equivalent
☐ Associate degree (occupational, technical, or vocational)
☐ College (bachelor's degree)
☐ Graduate school (masters, professional, or doctoral degree)
☐ Don't know
☐ Other_____

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EMPLOYMENT, RETIREMENT, OR DISABILITY

What is your current status? (Check one)

☐ Employed full-time (35 hours or more per week)

☐ Employed part-time (less than 35 hours per week)

☐ Homemaker

☐ Retired

☐ Unemployed (not due to disability)

☐ Unemployed (due to DM)

☐ Unemployed (due to another disability)

If employed, what is your current occupation?

☐ Job title: _____

Comments:_____

Has myotonic dystrophy affected your employment? ☐ Yes ☐ No

If yes, how was your job affected? (Check all that apply)

☐ Lost job ☐ Early retirement ☐ Forced to go on disability

☐ Job modified to accommodate physical limitations

ASSISTIVE DEVICES

	Check the box for any devices you have ever used	Age when you started use (Estimate if not sure)	Age when you stopped use (Estimate if not sure) *Leave blank if still using*
Ankle and/or knee braces	☐	Years old	Years old
Long leg braces	☐	Years old	Years old
Cane	☐	Years old	Years old
Walker	☐	Years old	Years old
Wheelchair or scooter (check one)	Long distances only ☐ Usually ☐ Always ☐	Years old	Years old
CPAP (Continuous Positive Airway Pressure)	☐	Years old	Years old
BiPAP (Bi-level Positive Airway Pressure)	☐	Years old	Years old
Oxygen therapy (at home)	☐	Years old	Years old
Ventilator	☐	Years old	Years old
Pacemaker and/or defibrillator	☐	Years old	Years old
Other	☐	Years old	Years old

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SIGNS AND SYMPTOMS

Does myotonia (cramping, difficulty releasing your grip, etc.) currently affect your normal daily activities? (Check one)

☐ Yes, severely

☐ Yes, moderately

☐ Yes, mildly

☐ No, not at all

	Check the box for any symptoms you have ever had	Age when symptom started (Estimate if not sure)	Age when symptom stopped (Estimate if not sure) *Leave blank if still present*
Trouble with hands/grip locking up, or hand stiffness	☐	Years old	Years old
Difficulty making a tight fist, loss of grip strength or difficulty opening jars	☐	Years old	Years old
Trouble speaking clearly	☐	Years old	Years old
Trouble with swallowing	☐	Years old	Years old
Weakness of face	☐		
Difficulty walking on toes or heels, or ankle weakness	☐	Years old	Years old
Difficulty getting up from the floor, rising from a chair, or climbing stairs	☐	Years old	Years old
Trouble with breathing or shortness of breath	☐	Years old	Years old
Cataracts	☐	Years old	Years old
Racing heartbeat, irregular heartbeat, palpitations	☐	Years old	Years old
Baldness or thinning hair	☐	Years old	Years old

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BROKEN BONES AND SURGERIES

Have you ever had a broken bone or surgery? ☐ Yes ☐ No

If yes, list them and the year they occurred. (*If you need more space, use back of form)*

Broken bones / Surgeries	**Year occurred**

MEDICATIONS / SUPPLEMENTS

Do you take medications or supplements? ☐ Yes ☐ No

If yes, please list prescriptions, over-the-counter medications, and supplements that you have taken in the past 2 months and why you take them. *(If you need more space, use the back of this form)*

Prescriptions, over the counter medications, and supplements	**Indication** (reasons why you are taking it)

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ALLERGIES List any food or drug allergies.

_____	_____	_____
_____	_____	_____
_____	_____	_____

TOBACCO (NICOTINE) USE

Do you use or have you ever used tobacco? Examples include cigarettes, chewing tobacco, pipes, or electronic nicotine delivery systems, like vaporizers or electronic cigarettes.

- ☐ Yes, I use tobacco currently (within the past 2 months)
- ☐ Yes, I used tobacco in the past (more than 2 months ago)
- ☐ No, I've never used tobacco

TREATMENT or COUNSELING

	Check the box for any treatments you have ever had	Age when you started treatment (Estimate if not sure)	Age when you stopped treatment (Estimate if not sure) *Leave blank if still receiving treatment*
Aquatic (water) therapy	☐	Years old	Years old
Emotional or psychological counseling	☐	Years old	Years old
Genetic counseling	☐	Years old	Years old
Occupational therapy	☐	Years old	Years old
Physical therapy	☐	Years old	Years old
Speech therapy	☐	Years old	Years old
Vocational rehabilitation	☐	Years old	Years old
Other:	☐	Years old	Years old

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OTHER MEDICAL PROBLEMS

	Check the box for any medical problems you have ever had	Check the box if the medical problem is ongoing
Acid reflux or "heartburn"	☐	☐
Asthma	☐	☐
Cancer or tumor	☐	☐
Type of cancer or tumor:		
Chronic infection	☐	☐
Constipation	☐	☐
Diabetes	☐	☐
Emphysema	☐	☐
Gallbladder trouble	☐	☐
Heart disease	☐	☐
High blood pressure	☐	☐
High cholesterol	☐	☐
Kidney trouble	☐	☐
Liver trouble	☐	☐
Miscarriage	☐	N/A
Prostate trouble	☐	☐
Psychological problem: depression / anxiety	☐	☐
Rheumatoid arthritis	☐	☐
Stillbirth	☐	N/A
Stroke	☐	☐
Stomach ulcers	☐	☐
Thyroid: high / hyperthyroidism	☐	☐
Thyroid: low / hypothyroidism	☐	☐
Thyroid: nodules	☐	☐
Trouble with sexual function	☐	☐
Other:	☐	☐
Other:	☐	☐

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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 = *slight* chance of dozing
2 = *moderate* chance of dozing
3 = *high* chance of dozing

Situation	Chance of dosing
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	

Participation in other research studies

Are you registered with any other myotonic dystrophy registry? ☐ Yes ☐ No ☐ Not sure

If yes, what is the name of the registry: _____

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

☐ Yes, multiple times ☐ Yes, once ☐ No

Have you ever received an experimental treatment for myotonic dystrophy? ☐ Yes ☐ No

If yes, what was that treatment: _____

Did anyone help fill out this form? If yes, who helped you?

Name of individual filling out the form: _____

Relationship to applicant: _____

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EMERGENCY CONTACT

Please provide the name, address, and telephone number of a person we can contact if needed (e.g., we don't have your current address or contact information). Please print.

Name: _____ Relationship: _____

Address: _____ City: _____ State: _____

Phone number: _____ Zip code: _____

This is the end for the form. Thank you for your support of the Registry.

The Registry is supported through the National Institute of Arthritis and Musculoskeletal and Skin Diseases and the National Institute of Neurological Disorders and Stroke (grant #U54-NS048843 and contracts #N01-AR-5-2274 and #NO1-AR-0-2250).

Contents of this form were made, in whole, or in part, by the following members of the **Scientific Advisory Committee** of the National Registry.

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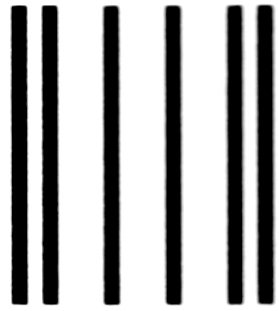

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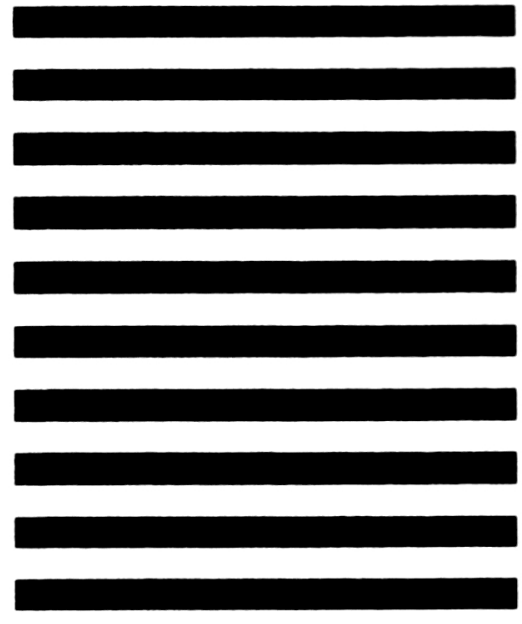

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