



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



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.
- You can choose to complete the forms for the Registry on paper or online.
- Procedures include completing a questionnaire at enrollment and then providing updates to your information once a 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.
- It will take you about 20-45 minutes to complete your enrollment and questionnaire. Then it will take about 15 minutes to update your information once a year. Your overall participation in this study will last for the next 5-10 years or longer.
- 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;

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Registry Number: _____

RSRB Approval Date: 8/20/2025
Expiration Date: 8/19/2026

- 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.

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 available to not-for-profit and academic centers and is a secure, web-based application for 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. You can skip any of the questions you do not want to answer.
- Participation of family members is strongly encouraged. Each family member is encouraged to enroll in the Registry and to complete the forms themselves, if interested and able. On your questionnaire, you can choose to share with us whether you have family members affected by myotonic dystrophy or FSHD, their names, and whether they are in the Registry or are planning to participate in the Registry. We will only record family relationships if both you and your family member agree to do so. No information about you will be shared with members of your family.

- **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. We will only request your records once. 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

- Learning about opportunities to participate in research studies or drug trials. 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. Before we inform you about such studies, 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. It is your choice whether you want to participate in a study. If you are interested to learn more about such studies, you can contact the researcher for more information or access the study information provided to you (e.g. online). **The Registry will not provide any information that could identify you (e.g. name, address etc.) to third parties.**

Annual update Questionnaire: 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. On your Patient Information Form or questionnaire, you can choose whether you would like to complete annual updates online or by postal mail. If you complete the form online, you will be emailed a link and asked to enter your age and last name to verify yourself to complete the update form.

- 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.
- 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.

If your child is enrolled in the Registry as a minor and turns 18 years old during follow-up, we will send you an updated consent form to review. Your child will have the opportunity to ask questions and sign and return the form if they would like to continue in the Registry as an adult. If the consent is not returned, we will send three reminder letters. If the consent form is not returned, they will no longer receive updates from the Registry, and the information already collected in the Registry will be retained.

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 include clarification or copies of the forms you completed, annual requests to update your information (example, annual forms sent by email with links to REDCap), general correspondence, announcements about opportunities to participate in other research (with links to more information or with contact information for other researchers), 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 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 <i>(initial)</i> | NO <i>(initial)</i> |
|--------------------------------|-------------------------------|

I consent to the use of **email** in this study. If yes, enter email address:

Share a copy of your genetic test results for DM or FSHD if available.

| | | |
|--------------------------------|-------------------------------|--|
| YES <i>(initial)</i> | NO <i>(initial)</i> | Have not had a genetic test <i>(initial)</i> |
|--------------------------------|-------------------------------|--|

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

| | |
|--------------------------------|-------------------------------|
| YES <i>(initial)</i> | NO <i>(initial)</i> |
|--------------------------------|-------------------------------|

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

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: _____



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 types of muscular dystrophy. The two muscular dystrophies 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 myotonic dystrophy and people with FSHD.
- To share information with doctors and scientists so that they can learn more about the cause of muscular dystrophy 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 muscular dystrophy 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 health, and symptoms or problems you may have. We may collect information from your doctor to learn more about your symptoms. You have the option to share some of your medical record with us. You can choose to complete the forms for the Registry on paper or online.

It will take you about 20-45 minutes to complete your enrollment and questionnaire. We will send you a new questionnaire each year to see if you have any changes (new address, new phone number, or new symptoms). These forms help us keep track of how symptoms change over time. Then it will take about 15 minutes to update your information once a year.

If you decide to join the Registry, you may be asked at a later time if you would like to help with other studies. 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.

When you turn 18 years old, we will send you an adult version of this consent form to see if you are still interested in participating and to answer any questions. If the consent is not returned, we will send you three reminder letters to get the updated form. If not returned after three attempts, you will no longer receive new information from us. Your information already collected will be retained in our database.

How long will you be in this study?

Your overall participation in this study will last for the next 5-10 years or longer.

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:

Ver 10: STUDY #00000010

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

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: _____

Below Completed by Registry Staff Only

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: _____

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

You have the option to share with us names of your family members who are in the Registry or planning to participate in the Registry. Please write their names below if you chose to.

| Name | Relationship to you | Name | Relationship to you |
|------|---------------------|------|---------------------|
| 1. | | 7. | |
| 2. | | 8. | |
| 3. | | 9. | |
| 4. | | 10. | |
| 5. | | 11. | |
| 6. | | 12. | |

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

Are you currently in school? Yes No

If yes, indicate the grade or level below:

- Attending elementary, middle, or high school (Circle one grade below):
 K 1 2 3 4 5 6 7 8 9 10 11 12
- Attending technical/professional school
- Attending college
- Attending graduate school

If no, indicate highest level of education completed below:

- No formal education
- High school, GED or equivalent
- Associate degree (occupational, technical, or vocational)
- College (bachelor’s degree)
- Graduate school (masters, professional, or doctoral degree)
- Other _____

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 <u>ever</u> used | Age when you <u>started</u> use (Estimate if not sure) | Age when you <u>stopped</u> use (Estimate if not sure) <i>Leave blank if still using</i> |
|--|---|--|---|
| Ankle and/or knee braces | <input type="checkbox"/> | Years old | Years old |
| Long leg braces | <input type="checkbox"/> | Years old | Years old |
| Cane | <input type="checkbox"/> | Years old | Years old |
| Walker | <input type="checkbox"/> | Years old | Years old |
| Wheelchair or scooter (check one) | Long distances only <input type="checkbox"/> | Years old | Years old |
| | Usually <input type="checkbox"/> | | |
| | Always <input type="checkbox"/> | | |
| CPAP (Continuous Positive Airway Pressure) | <input type="checkbox"/> | Years old | Years old |
| BiPAP (Bi-level Positive Airway Pressure) | <input type="checkbox"/> | Years old | Years old |
| Oxygen therapy (at home) | <input type="checkbox"/> | Years old | Years old |
| Ventilator | <input type="checkbox"/> | Years old | Years old |
| Pacemaker and/or defibrillator | <input type="checkbox"/> | Years old | Years old |
| Other | <input type="checkbox"/> | Years old | Years old |

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 <u>ever</u> had | Age when symptom <u>started</u> (Estimate if not sure) | Age when symptom <u>stopped</u> (Estimate if not sure) <i>Leave blank if still present</i> |
|--|---|---|--|
| Trouble with hands/grip locking up, or hand stiffness | <input type="checkbox"/> | Years old | Years old |
| Difficulty making a tight fist, loss of grip strength or difficulty opening jars | <input type="checkbox"/> | Years old | Years old |
| Trouble speaking clearly | <input type="checkbox"/> | Years old | Years old |
| Trouble with swallowing | <input type="checkbox"/> | Years old | Years old |
| Weakness of face | <input type="checkbox"/> | | |
| Difficulty walking on toes or heels, or ankle weakness | <input type="checkbox"/> | Years old | Years old |
| Difficulty getting up from the floor, rising from a chair, or climbing stairs | <input type="checkbox"/> | Years old | Years old |
| Trouble with breathing or shortness of breath | <input type="checkbox"/> | Years old | Years old |
| Cataracts | <input type="checkbox"/> | Years old | Years old |
| Racing heartbeat, irregular heartbeat, palpitations | <input type="checkbox"/> | Years old | Years old |
| Baldness or thinning hair | <input type="checkbox"/> | Years old | Years old |

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 |
|---------------------------------|----------------------|
| | |
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MEDICATIONS / SUPPLEMENTS

Do you take medications or supplements? Yes No Not sure

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 <u>ever</u> had | Age when you <u>started</u> treatment (Estimate if not sure) | Age when you <u>stopped</u> treatment (Estimate if not sure) <i>Leave blank if still receiving treatment</i> |
|---------------------------------------|---|--|---|
| Aquatic (water) therapy | <input type="checkbox"/> | Years old | Years old |
| Emotional or psychological counseling | <input type="checkbox"/> | Years old | Years old |
| Genetic counseling | <input type="checkbox"/> | Years old | Years old |
| Occupational therapy | <input type="checkbox"/> | Years old | Years old |
| Physical therapy | <input type="checkbox"/> | Years old | Years old |
| Speech therapy | <input type="checkbox"/> | Years old | Years old |
| Vocational rehabilitation | <input type="checkbox"/> | Years old | Years old |
| Other: | <input type="checkbox"/> | Years old | Years old |

OTHER MEDICAL PROBLEMS

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

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: _____

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: _____

ANNUAL UPDATE

Would you prefer to receive your annual questionnaire online or by postal mail (check one below)?

Online

Postal mail

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

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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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