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:

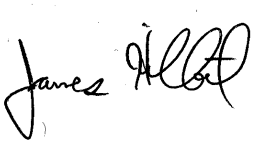
1. Consent Form - Please sign and return one copy. The second copy is for you to keep.
2. Assent Form – Completed if the enrollee is a child between the ages of 13-17 years old.
3. Patient Information Form

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: 8/20/2025

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;

Registry Number: _____

- 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

Registry Number: _____

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.

Registry Number: _____

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,

Registry Number: _____

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 *(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 results for DM or FSHD if available.

YES *(initial)*	NO *(initial)*	Have not had a genetic test *(initial)*

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

Registry Number: _____

2.) <u>CONSENT FROM PARENT, LEGAL GUARDIAN, or LEGALLY AUTHORIZED REPRESENTATIVE (LAR)</u>

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

<u>PERSON OBTAINING CONSENT</u>

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: _____

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.

Registry Number: _____

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

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:

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:_____

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

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

Patient Information Form

Facioscapulohumeral Muscular Dystrophy (FSHD)

Date: _____

Name: _____
First Middle Last (Maiden)

Address: _____
Street

_____
City State Zip Code

Telephone (list up to three numbers and circle which type):

Phone #: ____ - _____ - _____ (home, cell, work, or family)

Phone #: ____ - _____ - _____ (home, cell, work, or family)

Phone #: ____ - _____ - _____ (home, cell, work, or family)

Email Address: _____

Date of Birth: ____ / ____ / _____
Mo 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 ☐ Unknown

Race: (Check all that apply)
☐ American Indian or Alaskan Native ☐ Asian ☐ Black or African American
☐ Native Hawaiian or other Pacific Islander ☐ White ☐ Unknown

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

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

Where did you learn about this Registry?

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

FSHD ONSET AND DIAGNOSIS

What was the first symptom of FSHD? _____
_____

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

How old were you when your FSHD 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 FSHD ☐ Yes ☐ No ☐ Not sure

Who made your diagnosis of FSHD? (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 FSHD?

☐ 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.	

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

In the table below, indicate any blood relative, alive or deceased, that has or had FSHD.

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

	Had/has FSHD	**Unaffected**	**Unsure**
	Check appropriate boxes below		
Mother			
Father			
	Number with FSHD	**Number without FSHD**	**Number Unsure**
Grandparents			
Children < 18 years old			
Children ≥ 18 years old			
Grandchildren			
Siblings			
Half-Siblings			
Aunts or uncles			
Other (specify below):			

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 from the list below? (Check one)

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

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

☐ Homemaker

☐ Retired

☐ Unemployed (not due to disability)

☐ Unemployed (due to FSHD)

☐ Unemployed (due to another disability)

If employed, what is your current occupation?

☐ Job title:_____

Comments_____

Has FSHD affected your employment? ☐ Yes ☐ No

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

☐ Lost job

☐ Job modified to accommodate your physical limitations

☐ Forced to go on disability

☐ Early retirement

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 or hiking stick	☐	Years old	Years old
Abdominal brace	☐	Years old	Years old
Walker	☐	Years old	Years old
Wheelchair or scooter (check all that apply)			
	Long distances ☐	Years old	Years old
	Usually ☐	Years old	Years old
	Always ☐	Years old	Years old
Other	☐	Years old	Years old

SIGNS AND SYMPTOMS

Are you □ right or □ left handed?

Do you have any of the following?	**YES**	**NO**
1. Is one arm noticeably more affected by the disease?		
If yes, which is weaker: □ left □ right		
2. Is one leg noticeably more affected by the disease?		
If yes, which is weaker: □ left □ right		
3. Have you had surgery to fix your shoulder blades?		
If yes, which side: □ left □ right □ both		
4a. Do you have difficulty breathing?		
If yes, does your doctor feel it is related to your FSHD?		
4b. Do you require a breathing machine?		
If yes, which machine do you use: □ BiPAP □ CPAP □ Ventilator □ Other (type:)		
5. Have you had heart problems?		
If yes, what type: □ palpitations □ abnormal fast heart rate □ abnormal slow heart rate □ heart failure □ heart attack or angina		
6. Have you been diagnosed with hearing loss?		
Do you wear a hearing aid?		
7. Have you had any eye problems? (Other than needing glasses or contacts)		
If yes, check all that apply: □ retinal hemorrhage □ retinal detachment □ Coat's Disease □ other_____		
8. Do you have muscle or joint pain?		
If yes, check all areas that have pain: □ neck/upper back □ shoulder/upper arms □ lower back/hips □ elbows □ knees/thighs □ ankles/lower legs		

BROKEN BONES AND SURGERY

Have you ever had a broken bone or operation? □ Yes □ No

If yes, please list them and the year they occurred. *If you need more space on the paper form to write additional broken bones and surgeries, please use the back of this page or a separate sheet.*

Broken bone or surgery	**Year occurred**

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CURRENT ABILITIES AND RESTRICTIONS IN MOVEMENT

Facial Weakness:

Are your eyes occasionally dry and irritated?	☐ Yes	☐ No
Are your eyes always dry and irritated?	☐ Yes	☐ No
Do you have difficulty pronouncing certain words?	☐ Yes	☐ No
Do you have difficulty swallowing?	☐ Yes	☐ No
Do you have trouble whistling or drinking through a straw?	☐ Yes	☐ No

Arm Function: Which statement best describes your ability? (Check **one**)

Able to raise arms up sideways over head ☐

Able to raise arms sideways but not above shoulder level but do not need assistance for activities such as combing/shampooing hair, shaving, applying makeup, brushing teeth, etc. ☐

Able to raise arms sideways but not above shoulder level but do need assistance for activities such as combing/shampooing hair, shaving, applying makeup, brushing teeth, etc. ☐

Unable to raise arms sideways ☐

Leg Function: Which statements best describe your ability? (Check **all that apply**)

Able to walk and run ☐

Able to walk but not run ☐

Able to walk and climb stairs without using hand rail or cane ☐

Able to walk and climb stairs only with the help of railing or cane ☐

Able to walk with cane/walker but unable to climb stairs ☐

Unable to walk ☐

Mobility/Transfers: Which statement best describes your ability? (Check **one**)

When getting up from a chair, you:

Get up without using your arms (i.e., with arms folded across your chest) ☐

Need to use your arms to push up from the chair ☐

Use specific maneuvers to get up from a chair ☐

Get up only with the assistance of a person or device ☐

When getting out of bed, you:

Sit up from a lying position in bed without any problems ☐

Sit up from a lying position in bed only by using your arms ☐

Sit up from a lying position in bed only by turning sideways and using your arms ☐

Sit up from a lying position in bed only with someone's assistance ☐

Transfer from bed to chair only with assistive devices (ie: walker, bed rails) ☐

MEDICATIONS

Do you take medications or supplements? ☐ Yes ☐ No ☐ Not sure

If yes, please list any prescriptions, over the counter medications, and supplements you have taken in the past 2 months and why you take them. *If you need more space on the paper form to write additional medications, please use the back of this page or a separate sheet.*

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

ALLERGIES

List any food or drug allergies.

_____ _____

_____ _____

TOBACCO (NICOTINE) USE

Do you use or have you ever use tobacco? Example 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 have never used tobacco

TREATMENTS 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
Pneumonia	☐	☐
Prostate trouble	☐	☐
Psychological problem: depression / anxiety	☐	☐
Rheumatoid arthritis	☐	☐
Stillbirth	☐	N/A
Stomach ulcers	☐	☐
Stroke	☐	☐
Thyroid: high / hyperthyroidism	☐	☐
Thyroid: low / hypothyroidism	☐	☐
Thyroid: nodules	☐	☐
Trouble with sexual function	☐	☐
Other:	☐	☐
Other:	☐	☐

PARTICIPATION IN OTHER RESEARCH STUDIES

Are you enrolled in another FSHD registry? ☐ Yes ☐ No ☐ Not sure
If yes, what is the name of the registry: _____

Have you ever participated in a research study for FSHD?
☐ Yes, multiple times ☐ Yes, once ☐ No

Have you ever received an experimental treatment for FSHD? ☐ Yes ☐ No
If yes, what was that treatment:_____

ASSISTANCE COMPLETING THIS FORM

Did anyone help fill out this form? ☐ Yes ☐ No
If yes, list the name of individual filling out the form: _____
Relationship to applicant: _____

EMERGENCY CONTACT

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: _____ 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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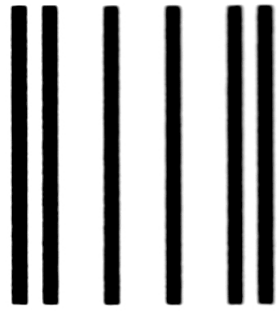

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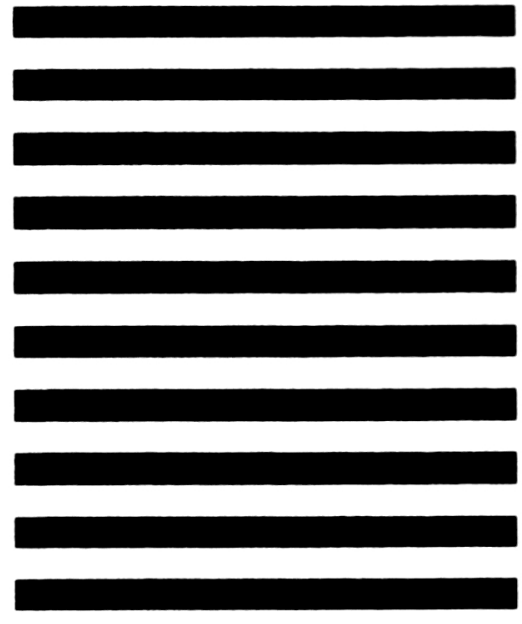

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