Connecting people with researchers

UNIVERSITY of ROCHESTER MEDICAL CENTER

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 (DM) and FSHD with researcher opportunities. Anyone with DM 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
4. Medical Information Form - This form gives us permission to request your medical records from your neurologist or primary care physician.

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   Elizabeth A. Luebbe

James Hilbert, MS
Health Project Coordinator

Elizabeth Luebbe
Health Project Coordinator

CONSENT FORM

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

Principal Investigator: Rabi Tawil, 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.

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.

- **Complete Authorization for Release of Medical Information form.** 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 will only request this information if you have received a genetic test or other exams that show that you do not have muscular dystrophy.

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 oremail 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

When using e-mail to communicate with you in this study, the researcher cannot guarantee, but will use reasonable means to maintain security and confidentiality of e-mail information sent and received. You and the researcher should understand the following conditions, instructions and risks of e-mail use:

Conditions for e-mail use:
a) E-mail is not appropriate for urgent or emergency situations. The researcher cannot guarantee that any particular e-mail will be read and responded to.
b) E-mail must be concise. You should schedule an appointment if the issue is too complex or sensitive to discuss via e-mail.
c) E-mail communications between you and the researcher will be filed in your research record.
d) Your messages may also be delegated to any member of the study team for response.
e) The researcher will not forward subject-identifiable e-mails outside of URMC and Affiliates without your prior written consent, except as authorized or required by law.
f) You should not use e-mail for communication regarding sensitive medical information.
g) It is your responsibility to follow up and/or schedule an appointment if warranted.

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

Risks of e-mail use:

Sending your information by e-mail has a number of risks that you should consider. These include, but are not limited to, the following:

a) E-mail can be circulated, forwarded, stored electronically and on paper, and broadcast to unintended recipients.

b) E-mail senders can easily misaddress an e-mail.
c) Backup copies of e-mail may exist even after the sender or the recipient has deleted his or her copy.
d) Employers and on-line services have a right to inspect e-mail transmitted through their systems.
e) E-mail can be intercepted, altered, forwarded, or used without authorization or detection.
f) E-mail can be used to introduce viruses into computer systems.

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.

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:____________________

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: Rabi Tawil, 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 will collect information from your doctor to learn more about your symptoms if you have a muscle problem. We will also collect information from your doctor if you had test that says you don't have a muscle problem.

RSRB Approval Date: 9/8/2022
Expiration Date: 9/7/2023

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 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 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 call us and we will remove your name. No one will be upset with you.

Will you get any money or gifts for being in this study?

You will not be paid or given anything for being in this study.

What if you have questions about this study?

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

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

What if you have questions about your rights as a research subject?

Please contact the University of Rochester Research Subjects Review Board at 265 Crittenden Blvd., CU 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: ______________

RSRB Approval Date: 9/8/2022
Expiration Date: 9/7/2023

UNIVERSITY of ROCHESTER MEDICAL CENTER

& Affiliates

University of Rochester Department of Neurology
National Registry for DM and FSHD
601 Elmwood Ave, Box 673
Rochester, NY 14642
Telephone: (888) 925-4302 Fax: (585-276-1947)

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

PLEASE PRINT.

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

Address: Patient's phone#:

City/State/Zip:

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

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

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

Name of Provider/ Person/Facility ______________________ Address ______________________

City, State, Zip Code ______________________ Phone #/Fax# include area code ______________________

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

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

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

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

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

- ☐ Treatment summary (includes discharge summary, history/physical, laboratory tests, x-ray reports, operative reports, pathology)
- ☐ Specific information or reports (describe): ______________________
- ☐ Other (describe): ______________________

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

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

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

☐ This request only

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

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

I understand that:

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

Signature of Patient or Representative ______________________________ Date ______________

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

Revised 8/11

Distribution. Original to medical record. Copy to patient as required
This authorization must be retained for a m1n1mum of six years beyond the validation limits

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

Patient Information Form *for individuals with Facioscapulohumeral Muscular Dystrophy(FSHD)*

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

Date: ____________________

NAME:____
First Middle (Maiden) Last

ADDRESS:____
Street

____
City State Zip Code

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

EMAIL ADDRESS: ____

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

Where did you learn about the Registry?

☐ Your doctor
☐ Family
☐ Friend
☐ Other____________________________________

☐ Internet
☐ Support group

☐ MDA
☐ Magazine/Newsletter

For office use only. Name: ______________________________________ Registry Number: ________________________

INFORMATION ABOUT YOUR DIAGNOSIS OF FSHD:

1. What was the first symptom of FSHD?______________________________________
____

2. How old were you when you had your first symptom of FSHD? (Give your best estimate even if you are not sure.)________years old.

3. How old were you when your FSHD was diagnosed? (Give your best estimate even if you are not sure.)________years old.

4. Did you have any of these tests?

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

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

6. Were you the first person in your family to have the diagnosis of FSHD?
   ☐ Yes ☐ No ☐ Not sure

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

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

OCCUPATION AND EMPLOYMENT

What is your current occupation (complete below)
☐ Employed (describe your job)____
☐ Homemaker ☐ Student ☐ Retired
☐ Disabled because of FSHD ☐ Disabled (not due to FSHD)
☐ Unemployed (not due to disability)
Comments____

Has FSHD affected your employment? ☐ Yes ☐ No
If yes, how (check boxes)
☐ Lost job ☐ Forced to go on disability
☐ Job modified to accommodate your physical limitations ☐ Early retirement

EDUCATION

Highest level of education completed: (check appropriate box)
☐ No formal education
☐ Grade school
☐ High school
☐ Technical school
☐ College
☐ Graduate school
☐ Other_________________________
☐ Don't know

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

For office use only. Name: ______________________________ Registry Number: ____________________

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		
4. Do you have difficulty breathing?		
If yes, does your doctor feel it is related to your FSHD?		
Do you require a breathing machine? **If yes,** which machine do you use: ☐ BiPAP ☐ CPAP ☐ Ventilator		
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 affected: ☐ 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 date they occurred. *If you need more room, please use an additional piece of paper.*

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

CURRENT ABILITIES AND RESTRICTIONS IN MOVEMENT

To help us understand your current abilities/difficulties, please grade yourself in the following:

Facial Weakness: (Check as many as apply)
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?
(Please check **only one** box.)
Are you able to raise your arms up sideways over your head? ☐
Are you able to raise your 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. ☐
Are you able to raise your 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?
(Please check **all that apply**.)
Walk and run ☐
Walk but not run ☐
Walk and climb stairs without using hand rail or cane ☐
Walk and climb stairs only with the help of railing or cane ☐
Walk with cane/walker but unable to climb stairs ☐
Unable to walk ☐

Mobility/Transfers: Which statement best describes your ability?
(Please check **only one** box.)
When getting up from a chair are you able to:
Get up without using your arms (ie; 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 ☐

Getting out of bed are you able to:
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) ☐

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MEDICATIONS

Do you take medications? ☐ Yes ☐ No ☐ Don’t know

If yes, please give the name of each medication. Include both prescription and non-prescription drugs and herbal remedies.

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

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

If you need more room, please use an additional piece of paper.

What is your current height:_______feet_____inches, and weight:_________pounds

ALLERGIES

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

___________________________________ ___________________________________
___________________________________ ___________________________________
___________________________________ ___________________________________
___________________________________ ___________________________________

Do you smoke tobacco? ☐ Yes ☐ No

TREATMENTS OR COUNSELING

Have you ever received any of the following?

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

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OTHER MEDICAL PROBLEMS

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

☐ Diabetes
☐ High blood pressure
☐ Asthma
☐ Rheumatoid arthritis
☐ Emphysema
☐ Pneumonia
☐ Heart disease or heart beat irregularity
☐ Cancer or tumor, type____________
☐ High cholesterol
☐ Miscarriage
☐ Stillbirth

☐ Stroke
☐ Kidney trouble
☐ Thyroid trouble
☐ Stomach ulcers
☐ Gall bladder trouble
☐ Prostate trouble
☐ Liver trouble
☐ Chronic infection
☐ Trouble with sexual function
☐ Acid reflux or “heartburn”
☐ Constipation

☐ Psychological problems such as depression or anxiety
☐ Other____

ETHNICITY/RACE

Are you Hispanic or Latino? ☐ Yes ☐ No

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

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

☐ Asian
☐ White

Have you ever participated in a research study for FSHD ? ☐ Yes ☐ No ☐ Not sure

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

If yes, what was that treatment:____

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

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

NAME: _______________________________________RELATIONSHIP: ________________
ADDRESS: ____
PHONE NUMBER: ____

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

IMPORTANT

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

Thank you for your help with the Registry.

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

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

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