Patient Self-Report *Registration Medical Questionnaire (II)* "The International Dysferlinopathy Registry"

To register with the International Dysferlinopathy Registry, you first have to read the *Information for Patients* form, complete and sign the *Registration and Consent Questionnaire (I)*, and then complete the *Registration Medical Questionnaire (II)*. Your doctor(s) will also be asked to complete the *Registration Medical Questionnaire (III)*. This document is the paper version of the *Registration Medical Questionnaire (II)* for the International Dysferlinopathy Registry.

For patients with a valid e-mail address (and internet access), we invite you to create your personal user account by registering on the International Dysferlinopathy Registry website (www.dysferlinregistry.org). By logging into your account, you will then be able to securely complete the online (electronic) version of this *Questionnaire II*. At anytime, you will also be able to download the pdf version of your filled in questionnaire. However, you can use this paper copy as an example to show the questionnaire to your doctor.

For patients without internet access or without a valid e-mail address, you can complete this paper version of *Questionnaire II* and send it, together with your correctly completed and signed *Registration and Consent Questionnaire (I)*, to the following address:
The International Dysferlinopathy Registry ; Inserm UMR 910 ; Aix-Marseille Université; 27 boulevard Jean Moulin ; 13385 Marseille Cedex 05 ; FRANCE.

If you are the patient affected by a dysferlinopathy and are 18 years of age or over, you can fill in and sign this questionnaire yourself. If you are younger than 18 years of age, but can understand this questionnaire, you can complete and sign the form yourself, but you will need to also include your parent's or guardian's personal information. If you are the parent or guardian of a patient who is too young to or otherwise not able to complete the questionnaire by him/herself, you can complete it yourself on behalf of the patient.

If you prefer not to answer a question, please tick the appropriate box "☐ I choose not to answer this question". If this option is not offered, it means that we need you to answer that question in order to confirm your registration with the International Dysferlinopathy Registry. Your answers to those questions marked by the UMD-DYSF label are anonymously transferred on the UMD-DYSF database web portal (see point 4.1 in your *Registration and Consent Questionnaire (I)*).

---

1. YOUR DIAGNOSIS

1.1 YOUR CURRENT CLINICAL DIAGNOSIS

1.1.1 What is your CURRENT clinical diagnosis, according to your physician? UMD-DYSF

- ☐ Limb Girdle Muscular Dystrophy Type 2B (LGMD2B)
- ☐ Limb Girdle Muscular Dystrophy (I don't know about the LGMD type)
- ☐ Miyoshi myopathy
- ☐ Proximodistal form of dysferlinopathy
- ☐ Distal Myopathy with Anterior Tibial Onset (DMAT)
- ☐ HyperCKemia
- ☐ I do not have any symptom, I am asymptomatic
- ☐ Other *(please specify)*: _____
- ☐ I am not sure or I don't know

1.1.2. At what age were you diagnosed?
I was diagnosed at age: _____

1.1.3 What is your year of birth? _____

1.2 ONSET OF YOUR DISEASE

1.2.1 What were your FIRST presenting symptom(s)? *(several answers are possible – please choose all that apply)*

- ☐ Weakness of the: *(several answers are possible – please choose all that apply)*
  - ☐ Upper leg
  - ☐ Lower leg
  - ☐ Upper arm
  - ☐ Lower arm
  - ☐ Hands
  - ☐ Other *(please specify)*: _____
- ☐ High CK (CPK) levels
- ☐ Cramps
- ☐ Stiffness
- ☐ Pain
- ☐ Other *(please specify)*: _____

1.2.2 Was there a difference between your right and left sides when your symptoms FIRST presented?

- ☐ Yes, the difference was: _____
- ☐ No

1.2.3 When were your FIRST presenting symptom(s)?

I first noticed symptom(s) at age: _____

1.2.4 How were your FIRST muscle problems diagnosed? *(several answers are possible – please choose all that apply)*

- ☐ The problems were picked up because I have an affected relative
- ☐ The problems were picked up because of a coincidental blood test
- ☐ I had problems with my walking
- ☐ I had problems standing on my toes
- ☐ Other *(please specify)*: _____

1.3 YOUR PAST PHYSICAL ACTIVITY

Did you frequently participate in sports or other physical activity before your muscle weakness appeared?

- ☐ I choose not to answer this question
- ☐ No
- ☐ Yes. Please provide details about the type of your past sporting activities (eg, running, figure skating, ballet, American football, soccer, basketball, handball, rugby, tennis, swimming, etc), their weekly frequency and how many years you participated in them, and indicate at what age muscle problems began to interfere with each activity. *(several answers are possible – please choose all that apply)*

I have participated in: _____	_____ times a week	during _____ years	I noticed problems since the age of: ____
I have participated in: _____	_____ times a week	during _____ years	I noticed problems since the age of: ____
I have participated in: _____	_____ times a week	during _____ years	I noticed problems since the age of: ____

1.4 YOUR FAMILY BACKGROUND

Do members of your family have similar symptoms or the same diagnosis?

- ☐ Not to my knowledge
- ☐ I am not sure or I don't know
- ☐ I choose not to answer this question
- ☐ Yes I have a family member with similar symptoms or the same diagnosis.

Please detail your family relationship with your relative(s) (your mother, your father, a sister or a brother from the same two parents, a child, other relative(s)/ancestor(s)), and please specify -when you are aware of it- whether any of them has already joined this registry:

		Has this person already joined this registry?		
	Family relationship	Yes	No	I am not sure or I don't know
☐	Mother	☐	☐	☐
☐	Father	☐	☐	☐
☐	A sister (from the same two parents)	☐	☐	☐
☐	A second sister (from the same two parents)	☐	☐	☐
☐	A brother (from the same two parents)	☐	☐	☐
☐	A second brother (from the same two parents)	☐	☐	☐
☐	A child	☐	☐	☐
☐	Another child	☐	☐	☐
☐	Another child	☐	☐	☐
☐	Another relative/ancestor*(please specify)*: _____	☐	☐	☐
☐	Another relative/ancestor*(please specify)*: _____	☐	☐	☐

1.5 YOUR MUTATIONAL ANALYSIS OF THE DYSFERLIN GENE UMD-DYSF

Your diagnosis included dysferlin mutational analysis that confirmed you as a carrier of one or more mutation(s) in the dysferlin gene (by DNA sequencing). Please indicate whether:

- ☐ A single disease-causing mutation was identified
- ☐ Two or more disease-causing mutations were identified
- ☐ I am not sure/I don't know how many disease-causing mutations were identified

It is very important that your dysferlin mutational information is entered correctly in this registry. Therefore, before confirming your registration, the curator of this registry will retrieve your genetic test result from your doctor (or medical centre / lab) that you have indicated in your *Questionnaire I.* Your genetic information will then be anonymously coded and transferred to the UMD-DYSF web portal.

1.6 YOUR BIOLOGICAL ANALYSIS OF THE DYSFERLIN PROTEIN UMD-DYSF

It is very important that your dysferlin biological information is entered correctly in this registry. Therefore, before confirming your registration, the curator of this registry will retrieve your protein level test result from your doctor (or medical centre / lab) that you have indicated in your *Questionnaire I.* Your protein information will then be anonymously coded and transferred to the UMD-DYSF web portal.

1.7 YOUR BIOLOGICAL ANALYSIS OF THE CREATINE KINASE PROTEIN

Measuring the level of creatine kinase (CK) in the blood can help doctors determine which tissue has been damaged. CK is also known as creatine phosphokinase (CPK) and its level can be measured by a standard blood test. The curator of this registry will retrieve your CK level test result from your doctor (or medical centre / lab) that you have indicated in your *Questionnaire I.*

2. YOUR SYMPTOMS

2.1 YOUR CURRENT MOTOR FUNCTION

Motor function describes a person's ability to move their body. For each question, please tick the box with the most appropriate answer. If you have always been able to perform the motor function, please tick the "always" box.

2.1.1 What is the best motor function you are CURRENTLY able to achieve among the following ones?

☐ I can run		
☐ I can walk unaided (without support of an assistive device)		
☐ I can walk with an assistive device (walker, brace, cane, etc) *(go to question 2.1.3)*		
☐ I cannot walk (*go to question 2.1.3)*	☐ since the age of: _____	☐ always

2.1.2 How far can you CURRENTLY walk without assistance?

☐ A long distance (e.g. more than a mile or two kilometres)		
☐ A medium distance (e.g. less than a mile/two kilometres, but more than a block/100 metres)	☐ I cannot walk further than this since the age of: _____	☐ always
☐ A short distance (e.g. a block or 100 metres/yards)	☐ I cannot walk further than this since the age of: _____	☐ always
☐ Across a room (e.g. 10 metres/yards)	☐ I cannot walk further than this since the age of: _____	☐ always
☐ A few steps	☐ I cannot walk further than this since the age of: _____	☐ always
☐ I cannot walk without assistance at all	☐ since the age of: _____	☐ always

2.1.3 If you CURRENTLY use an assistive device for walking or instead of walking, please indicate what kind of assistive device you use, at what age you started to use it, and how often you use it for moving. *(several answers are possible – please choose all that apply)*

☐ I do not use any assistive device for walking or instead of walking			
☐ I use a wheelchair/motorised scooter	☐ part-time	☐ all the time	I started at age: _____
☐ I use a walker/walking frame	☐ part-time	☐ all the time	I started at age: _____
☐ I use a cane/stick	☐ part-time	☐ all the time	I started at age: _____
☐ I use long leg braces/callipers	☐ part-time	☐ all the time	I started at age: _____
☐ I use below knee splints/braces/AFO's	☐ part-time	☐ all the time	I started at age: _____
☐ I use other assistive device *(please specify)* _____	☐ part-time	☐ all the time	I started at age: _____

2.1.4 What is the best motor function you are CURRENTLY able to achieve when climbing four or more stairs?

☐ I can climb the stairs easily and independently (without leaning on the wall or using the railing, without an assistive device for walking and without help from another person)		
☐ I can climb the stairs independently (without leaning on the wall or using the railing, without an assistive device for walking and without help from another person), but with difficulty	☐ since the age of: ____	☐ always
☐ I can climb the stairs by myself, but I need to lean on the wall or use the railing or an assistive device for walking	☐ since the age of: ____	☐ always
☐ I can climb the stairs, but with help from another person	☐ since the age of: ____	☐ always
☐ I cannot climb stairs at all	☐ since the age of: ____	☐ always

2.1.5 What is the best motor function you are CURRENTLY able to achieve when descending four or more stairs?

☐ I can descend the stairs easily and independently (without leaning on the wall or using the railing, without an assistive device for walking and without help from another person)		
☐ I can descend the stairs independently (without leaning on the wall or using the railing, without an assistive device for walking and without help from another person), but with difficulty	☐ since the age of: ____	☐ always
☐ I can descend the stairs by myself, but I need to lean on the wall or use the railing or an assistive device for walking	☐ since the age of: ____	☐ always
☐ I can descend the stairs, but with help from another person	☐ since the age of: ____	☐ always
☐ I cannot descend stairs at all	☐ since the age of: ____	☐ always

2.1.6 What is the best motor function you are CURRENTLY able to achieve when standing up from a seated position?

☐ I can stand up easily and independently		
☐ I can stand up independently, but with difficulty	☐ since the age of: ____	☐ always
☐ I can stand up, but with the use of furniture or other assistive device	☐ since the age of: ____	☐ always
☐ I can stand up, but with help from another person	☐ since the age of: ____	☐ always
☐ I cannot stand up	☐ since the age of: ____	☐ always

2.1.7 What is the best motor function you are CURRENTLY able to achieve when sitting up from a lying position?

☐ I can sit up easily and independently		
☐ I can sit up independently, but with difficulty	☐ since the age of: ____	☐ always
☐ I can sit up, but with the use of furniture or other assistive device	☐ since the age of: ____	☐ always
☐ I can sit up, but with help from another person	☐ since the age of: ____	☐ always
☐ I cannot sit up	☐ since the age of: ____	☐ always

2.1.8 How long can you CURRENTLY stand without any support?

☐ More than 30 minutes		
☐ 10-30 minutes	☐ no more since the age of: ____	☐ always
☐ 5-10 minutes	☐ no more since the age of: ____	☐ always
☐ 1-5 minutes	☐ no more since the age of: ____	☐ always
☐ 1-10 seconds	☐ no more since the age of: ____	☐ always
☐ Not at all	☐ since the age of: ____	☐ always

2.1.9 Please indicate which tasks are CURRENTLY difficult or impossible for you to achieve without an assistive device:
(several answers are possible – please choose all that apply)

Walking on a slight elevation (like a ramp)	☐ I have no trouble	☐ Yes, I have trouble; since the age of: ____	☐ I can not perform this movement; since the age of: ____
Standing on tiptoes	☐ I have no trouble	☐ Yes, I have trouble; since the age of: ____	☐ I can not perform this movement; since the age of: ____
Raising your arms above your head	☐ I have no trouble	☐ Yes, I have trouble; since the age of: ____	☐ I can not perform this movement; since the age of: ____
Picking up a glass of water	☐ I have no trouble	☐ Yes, I have trouble; since the age of: ____	☐ I can not perform this movement; since the age of: ____
Unscrewing a jar	☐ I have no trouble	☐ Yes, I have trouble; since the age of: ____	☐ I can not perform this movement; since the age of: ____
Carrying a jug of water	☐ I have no trouble	☐ Yes, I have trouble; since the age of: ____	☐ I can not perform this movement; since the age of: ____
Turning the wheel of a car	☐ I have no trouble	☐ Yes, I have trouble; since the age of: ____	☐ I can not perform this movement; since the age of: ____
Typing (on a keyboard)	☐ I have no trouble	☐ Yes, I have trouble; since the age of: ____	☐ I can not perform this movement; since the age of: ____

2.1.10 Have you noticed any factors (e.g. patterns of exercise, diet, alcohol, others) that improve your motor ability or make your motor ability worse?

☐ Yes *(Please explain)*: _____

☐ No

2.2 YOUR CARDIAC AND RESPIRATORY FUNCTION

2.2.1 Have you been diagnosed with a heart condition?

☐ Yes

Please specify the type of heart condition you have been diagnosed with:

_____

☐ I am not sure or I don't know the type of heart condition I have been diagnosed with

☐ No

☐ I am not sure or I don't know

☐ I choose not to answer this question

2.2.2 Are you taking any medication for a heart condition?

☐ Yes

Please specify the heart medication you are taking: _____

☐ I am not sure or I don't know about the type of heart medication I am taking

☐ No

☐ I am not sure or I don't know

☐ I choose not to answer this question

2.2.3 To support their breathing, some muscular dystrophy patients get a ventilation device that they have to use either all day and night (all the time) or only a few hours per day or night (part-time). "Non-invasive ventilation" means that the patient uses the ventilation device without having had an operation (usually, this means that he/she wears a mask that can be removed at any time). "Invasive ventilation" means that the patient had to have an operation (a tracheotomy or a incision in the wind-pipe) to use the ventilation device. Do you use a ventilation device?

☐ Yes

☐ I use a non-invasive ventilation device	☐ part-time	☐ all the time
☐ I use an invasive ventilation device	☐ part-time	☐ all the time

☐ No
☐ I am not sure or I don't know
☐ I choose not to answer this question

2.2.4 To monitor your breathing function, your doctor might have done pulmonary function tests. One of the parameters measured in these tests is known as the Forced Vital Capacity (FCV). To test the FCV, the patients have to breathe in as far as they can and then blow out into a machine that measures how much air is being exhaled. The FCV is the volume of air exhaled and measured in litres. If you had this test done and know its result, please indicate that here.

☐ Yes, I had the FCV test done,

the result was:	_____(volume in litres)	☐ I am not sure or I don't know
on year:	_____	☐ I am not sure or I don't know

☐ No, I did not have the FCV test done
☐ I am not sure or I don't know whether or not I had the FCV test done
☐ I choose not to answer this question

2.3 YOUR MEDICAL TREATMENT

2.3.1 Have you ever received steroids (glucocorticoids) for your muscle problems?

☐ Yes
- ☐ I am currently on steroids, for _____ months.
- ☐ I was on steroids in the past. *Please specify the period(s) during which you were on steroids and your age at the beginning of each period:*
  At age _____ for _____ months
  At age _____ for _____ months
  At age _____ for _____ months

☐ No (go to question 2.3.3)
☐ I am not sure or I don't know (go to question 2.3.3)
☐ I choose not to answer this question (go to question 2.3.3)

2.3.2 If you are currently on steroids or were in the past, did you notice any improvement or decline on the steroids?

☐ Yes, I have noticed an improvement. *Please describe what kind of improvement you noticed:*

_____

☐ Yes, I have noticed a decline. *Please describe what kind of decline you noticed:*

_____

☐ No, I haven't noticed any improvement or decline
☐ I am not sure or I don't know

2.3.3 Please list all other medication, including vitamins and over the counter supplements that you are CURRENTLY taking:

_____
_____
_____
_____
_____
_____

☐ I do not take any other medication
☐ I choose not to answer this question

3. ADDITIONAL INFORMATION

3.1 What are your country and state/region of residence?
Country: _____
State/region: _____

3.2 What is the date of your last follow up?
Year: _____ Month: _____
☐ I choose not to answer this question

3.3 Are you currently involved in a clinical trial or research study regarding your muscle problem, or have been in the past? This information is of high relevance in the planning phase of a new trial or study!
☐ Yes, currently *(please specify)*: _____
☐ No but previously *(please specify):* _____
☐ No, I have never been involved in a clinical trial or research study
☐ I am not sure or I don't know

Patient's last/family name: Patient's first/given name:

_____ _____

Patient's date of birth: Day: _____ Month: _____ Year: _____

Patient's signature: Date:

_____ _____

Parent or guardian's name and signature: Date:

_____ _____

Please send this *Registration Medical Questionnaire (II)*, correctly completed and signed to: The International Dysferlinopathy Registry, Inserm UMR 910, Aix-Marseille Université, 27 boulevard Jean Moulin, 13385 Marseille Cedex 05, FRANCE.