Registration Medical Questionnaire (III) for the Medical Doctor "The International Dysferlinopathy Registry"

_____	_____	Born on _____.
Patient's First Name	Patient's Last Name	DD/MM/YYYY

Filled in by Dr. / Prof. _____
Doctor's Name

◘ This patient is my patient.
◘ This patient is NOT my patient (*please check the box and send this blank questionnaire back to the registry*).

1. CURRENT CLINICAL DIAGNOSIS

Please indicate the current clinical diagnosis for this patient:

- ◘ Limb Girdle Muscular Dystrophy Type 2B (LGMD2B)
- ◘ Limb Girdle Muscular Dystrophy (undetermined LGMD type)
- ◘ Miyoshi myopathy
- ◘ Proximodistal form of dysferlinopathy
- ◘ Distal Myopathy with Anterior Tibial onset (DMAT)
- ◘ HyperCKemia
- ◘ Asymptomatic
- ◘ Other *(please specify)*: _____

2. MUTATIONAL ANALYSIS OF THE DYSFERLIN GENE

◘ I DO NOT have any result of mutational analyses of the dysferlin gene for this patient.

Please note that the registry website provides both, a list of laboratories offering tests for the clinical diagnosis of dysferlinopathies and a list of laboratories performing genetic testing for dysferlinopathies. The Jain Foundation (www.jain-foundation.org) can also help guide physicians with patients with an unclear diagnosis.

◘ I DO have results of mutational analyses of the dysferlin gene for this patient.

Please note that information in this section is crucial for the registry. You can choose to fill in this section yourself or to send us a copy of the genetic report for this patient. If you choose to send the genetic report, please make sure that the report contains all the information required below.

To help you answer this section, you can consult the International Dysferlinopathy Registry website ("Links and Resources for Research and Medical Doctors") and/or the Human Genome Variation Society guidelines (www.hgvs.org/). Please enter disease-causing mutations only. Examples of annotating mutations in the standard HGVS nomenclature are: c.855+1delG; c.2997G>T; c.4872_4876delinsCCCC, etc. If the sequence of reference is not the one indicated below, please specify the accession number of the sequence of reference used to describe the variant. It is also very important for the registry to know where the analysis was performed.

MUTATION #1

Dysferlin genetic report attached ◘ Yes ◘ No Date of analysis: Year: _____ Month: _____

Medical centre / lab where the analysis was performed: _____

Mutation nomenclature at the DNA level *(following hgvs guidelines)*: _____

Mutation nomenclature at the protein level *(following hgvs guidelines)*: _____

Sequence of reference was: ◘ NM_ 003494.2 ◘ other (*please specify*):_____

This mutation was detected at: ◘ a homozygous state ◘ a heterozygous state

Detection method was: _____

MUTATION #2

Dysferlin genetic report attached ◘ Yes ◘ No Date of analysis: Year: _____ Month: _____

Medical centre / lab where the analysis was performed: _____

Mutation nomenclature at the DNA level *(following hgvs guidelines)*: _____

Mutation nomenclature at the protein level *(following hgvs guidelines)*: _____

Sequence of reference was: ◘ NM_ 003494.2 ◘ other (*please specify*):_____

This mutation was detected at: ◘ a homozygous state ◘ a heterozygous state

Detection method was: _____

MUTATION #3

Dysferlin genetic report attached ◘ Yes ◘ No Date of analysis: Year: _____ Month: _____

Medical centre / lab where the analysis was performed: _____

Mutation nomenclature at the DNA level *(following hgvs guidelines)*: _____

Mutation nomenclature at the protein level *(following hgvs guidelines)*: _____

Sequence of reference was: ◘ NM_ 003494.2 ◘ other (*please specify*):_____

This mutation was detected at: ◘ a homozygous state ◘ a heterozygous state

Detection method was: _____

3. BIOLOGICAL ANALYSIS OF THE DYSFERLIN PROTEIN

◘ I DO NOT have any result of biological analyses of the dysferlin protein for this patient.

◘ I DO have results of biological analyses of the dysferlin protein for this patient.

You can choose to fill in this section yourself or to send us a copy of the dysferlin protein biological report for this patient. If you choose to send a report, please make sure that the report contains all the information required below.

PROTEIN ANALYSIS RESULT #1

Dysferlin protein analysis report attached ◘ Yes ◘ No Date of analysis: Year: _____ Month: _____

Medical centre / lab where the analysis was performed: _____

Level of dysferlin: _____

Tissue: ◘ biopsy ◘ blood monocytes ◘ other *(please specify)*:_____

Detection method was: ◘ immunohistochemistry ◘ western blot ◘ other *(please specify)*:_____

PROTEIN ANALYSIS RESULT #2

Dysferlin protein analysis report attached ◘ Yes ◘ No Date of analysis: Year: _____ Month: _____

Medical centre / lab where the analysis was performed: _____

Level of dysferlin: _____

Tissue: ◘ biopsy ◘ blood monocytes ◘ other *(please specify)*:_____

Detection method was: ◘ immunohistochemistry ◘ western blot ◘ other *(please specify)*:_____

4. BIOLOGICAL ANALYSIS OF THE CREATINE KINASE PROTEIN

◘ I DO NOT have any result of biological analyses of the creatine kinase protein for this patient.

◘ I DO have results of biological analyses of the creatine kinase protein for this patient.

You can choose to fill in this section yourself or to send us a copy of the CK protein biological report for this patient. If you choose to send a report, please make sure that the report contains all the information required below.

Level of CK in blood:_____ IU/L , Year : _____ Month :_____ CK report attached ◘ Yes ◘ No

Level of CK in blood:_____ IU/L , Year : _____ Month :_____ CK report attached ◘ Yes ◘ No

Level of CK in blood:_____ IU/L , Year : _____ Month :_____ CK report attached ◘ Yes ◘ No

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Please send this *Registration Medical Questionnaire (III)* correctly filled in by e-mail (contact@dysferlinregistry.org), or by post to: The International Dysferlinopathy Registry; Inserm UMR S910, Aix-Marseille Université, 27 boulevard Jean Moulin, 13385 Marseille Cedex 05, FRANCE.