Information for Medical Doctors
"The International Dysferlinopathy Registry"

1. "Why create a patient registry for dysferlinopathies?"

Given the rarity of dysferlinopathies, the difficulty of identifying enough eligible patients can lead to a significant delay in or halt the planning of scientific studies or clinical trials. The best way of ensuring that a sufficient number of suitable patients can be quickly found, is to make sure that patients' details are collected together in a single database or "patient registry" that contains the information that researchers or doctors will need, including each patient's particular genetic defect and other key information about their disease.

The International Dysferlinopathy Registry contains accurate and updated information about the patients' genetic mutations and clinical conditions. This patient registry is meant to help in the recruitment of patients for the "International Clinical Outcome Study for Dysferlinopathy", which entered the preparation phase in July 2011 and is anticipated to begin Fall of 2012 in centres in Europe, as well as for future scientific research studies or clinical trials (look for up-to-date information on the registry website: www.dysferlinregistry.org). A registry for dysferlinopathies can also support other activities to improve patient care, such as the assessment and dissemination of standards of care.

2. "Which patients are eligible to register with the International Dysferlinopathy Registry?"

This patient registry is specifically developed for patients affected with a dysferlinopathy, including the most frequent clinical presentations called Limb Girdle Muscular Dystrophy type 2B (LGMD2B) and Miyoshi myopathy, but also all other clinical presentations related to genetic defects in the gene called "dysferlin". This registry is exclusively designed for patients with a diagnosis of dysferlinopathy that is confirmed by a genetic test indicating that the patient carries one or more (predicted) pathogenic dysferlin mutation(s).

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.

3. "What are the different forms that my patient and I have to complete?"

To register with the International Dysferlinopathy Registry, your patient should first read the *Information for Patients* document carefully. If your patient decides to take part in this registry, he/she is asked to create a personal user account on the International Dysferlinopathy Registry website by completing and validating the online (electronic) version of his/her *Registration and Consent Questionnaire (I)*. By logging into his/her account, your patient will then be able to securely complete his/her online (electronic) *Registration Medical Questionnaire (II)*, with your help if necessary.

For patients without internet access, they will need to complete and sign paper copies of *Questionnaires I* and *II* and send them to the registry. Copies of all the registration forms can be easily downloaded from the registry website or sent by postal mail upon request.

By (electronically) validating or by signing their *Questionnaire I*, patients authorize the International Dysferlinopathy Registry to:

> *"(...) a) contact the doctor(s) / medical institution(s) that [they] indicated in this questionnaire in order to obtain the genetic and other medical information about [their] dysferlinopathy that are requested by the registry;*
> *b) store all [their] contact details and medical data – that have been provided by [them] or by the above doctor(s) / medical institution(s) – in this registry, where they may be used for research and for the planning of clinical trials and research studies; (…)"*

If you allow us to do so (by signing the *Consent for Doctor* form), we will add your contact information to the list of doctors/medical centres on the online *Questionnaire I*, from which the patients choose their health professional. In addition, we have attached a *Recruitment Leaflet* about the registry and would request that you put the same contact details on the back of this *Leaflet* and send it to all your genetically confirmed dysferlinopathy patients (i.e. patients who have at least one pathogenic mutation in the dysferlin gene) in order to encourage them to register with the International Dysferlinopathy Registry. Additional paper copies of *Recruitment Leaflets* can be requested by contacting the registry staff (see section 6) or a pdf version of the *Recruitment Leaflet* is available on the registry website. We feel that doing this will simplify and streamline the process for not only you, but also the patients and the registry staff.

Each time a new patient registers with the International Dysferlinopathy Registry and indicates you as his/her doctor/medical centre, we will contact you to obtain additional medical (genetic/biological/clinical) data for this patient. The medical data that we will ask you to provide – after obtaining full consent from your patient (see above) – include a copy of the patient's dysferlin genetic report and results of biological analyses of the patient's dysferlin and creatine kinase protein levels. To provide these data, we will ask you to fill in the *Registration Medical Questionnaire (III)* (see the attached example or the pdf version available on the registry website) and to send it back to us by the most convenient way for you (see contact details below).

Please note that your patient's registration with the International Dysferlinopathy Registry will only be confirmed after the registry receives his/her electronic or paper *Questionnaires I* and *II* correctly filled in and signed/validated, and the *Questionnaire III* correctly filled in and signed by you.

4. "Why do you ask health professionals to provide their patients' data to the International Dysferlinopathy Registry?"

For the International Dysferlinopathy Registry, it is essential that medical information be provided both by the patient and by professionals involved in the patient's care – after obtaining full consent by the patient (see above).

For research into treatments for dysferlinopathies, it is important that the researchers have precise information about the patients' dysferlin genetic mutations and about other relevant medical data. Since some of the patients' medical data, such as the genetic test results and protein level data, are complex and might not be available to the patients in the form of reports, such information needs to be provided by their doctors.

5. "Who will have access to the patient data stored in the International Dysferlinopathy Registry?"

All the data collected for the International Dysferlinopathy Registry will be stored in a secure manner, and all the patients' records will be de-identified and assigned a unique and anonymous code. Only the person in charge of the International Dysferlinopathy Registry or a person explicitly appointed by him/her will be able to "de-code" this anonymous identifier to get access to all the stored patients' personal details. The Jain Foundation could also have access to all the patients' data, but only if the patients provided specific consent for the registry to share their data with the Jain Foundation.

In anonymous form, the patients' medical data will be available to approved third parties around the world, with the aim to accelerate research into dysferlinopathies. Third-party access to the anonymous disease-related data will occur only after their data request is granted by an oversight committee. The data will be provided in the form of a data-analysis report.

In anonymous form, the patients' data related to the clinical diagnosis (e.g. LGMD2B, Miyoshi myopathy), results from the mutational analysis of the dysferlin gene, and results from the biological analysis of the dysferlin protein will be made publically available on the web portal of the UMD-DYSF mutation database (www.umd.be/DYSF/). UMD-DYSF is a "Locus Specific Database" that has been specifically created to compile and analyse up-to-date information about pathogenic mutations identified in the dysferlin gene and in patients diagnosed with a dysferlinopathy. To learn more about mutation records in the UMD-DYSF database, please visit the International Dysferlinopathy Registry website.

6. Contact information

If you would like any additional information about this registry, additional *Recruitment Leaflets*, or if you want your contact information to be withdrawn from this registry, please send your request to:

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Thank you for reading this doctor information sheet.