Contact details for the International Dysferlinopathy Registry

The International Dysferlinopathy Registry
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13385 Marseille Cedex 05
FRANCE

Website: www.dysferlinregistry.org

E-mail: contact@dysferlinregistry.org

Registry principal investigator: Dr. Martin Krahn

Contact details of the doctor to indicate in my *Registration Questionnaire*:

Sponsors and Partners

JAIN FOUNDATION

AFM Association Française contre les Myopathies

Inserm

Aix*Marseille université

APHM Assistance Publique Hôpitaux de Marseille

TREAT-NMD Neuromuscular Network

The International Dysferlinopathy Registry was initiated by and is funded in part by the Jain Foundation (contact@jain-foundation.org).

An International Clinical Outcome Study for Dysferlinopathy

The "Clinical Outcome Study for Dysferlinopathy" is anticipated to begin in mid-2012 in centres in Europe (UK, Spain, Germany, Italy and France), USA, Japan and Australia. The aim of this "Clinical Outcome Study" is to determine the clinical outcome measures required for future clinical trials, characterize the disease progression of dysferlinopathy and collect biological samples for the identification of disease markers that are needed to non-invasively monitor the disease during clinical trials. Without this information, effective clinical trials cannot be performed.

This study is recruiting a large number of genetically confirmed dysferlinopathy patients aged 10 years or older, who are ambulant or non-ambulant. Participants will be assessed at 6 visits over 3 years via medical, physiotherapy, and MRI/MRS assessments, as well as standard blood tests. Optionally, the participants can donate blood samples and a skin sample for use in the identification of disease markers and other approved research.

Recruitment is anticipated to start in mid-2012. Please note that funds are available to cover travel costs to the study centres.

Members of the International Dysferlinopathy Registry will receive updates regarding the Clinical Outcome Study for Dysferlinopathy.

To obtain additional information about or sign up for the Clinical Outcome Study, please contact one of the following:

- The nearest study centre - a list of centres is available via the study website (www.dysferlinoutcomestudy.org) or the International Dysferlinopathy Registry website (www.dysferlinregistry.org).
- The study coordinator at contact@dysferlinoutcomestudy.org.
- The Jain Foundation at contact@jain-foundation.org.

Oversight and funding for this study is provided by the Jain Foundation.

The principal investigator of the study is:
Prof. Kate Bushby, Institute of Genetic Medicine, Newcastle University, International Centre for Life, Central Parkway, Newcastle upon Tyne, NE1 3BZ, United Kingdom.

JAIN FOUNDATION

ORCHESTRATING A CURE
LGMD2B DYSFERLINOPATHY MIYOSHI
www.jain-foundation.org

The International Dysferlinopathy Registry

An opportunity for patients with LGMD2B, Miyoshi myopathy and other clinical presentations of dysferlinopathies to participate in research studies or clinical trials and obtain the best possible care

Register online at www.dysferlinregistry.org

Why is a patient registry for dysferlinopathies important?

Dysferlinopathies are very rare diseases affecting only 2-5 people per million. Given this rarity, the difficulty in identifying enough eligible patients can lead to significant delays in scientific studies or clinical trials. A registry ensures that a sufficient number of suitable patients are quickly found.

In addition, the **International Dysferlinopathy Registry** will provide key details and information needed by physicians and researchers, including the specific genetic defects found in each patient.

- The Registry helps identify potential participants for clinical trials and research studies, such as the International Clinical Outcome Study for Dysferlinopathy detailed on the back of this leaflet.
- The Registry helps researchers answer questions such as "How common are dysferlinopathies?" or "What are the precise genetic defects in dysferlinopathies?" This in turn helps raise awareness, improve diagnostics and increase research in dysferlinopathies.
- The Registry supports other activities to improve patient care, such as the assessment and dissemination of standards of care.

Who can register?

This patient registry is uniquely developed for all patients worldwide 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 dysferlin gene.

Please register if your dysferlinopathy diagnosis has been confirmed by a genetic test, verifying that you have one or more genetic defects (mutations) in the dysferlin gene.

If you are not sure whether a genetic test has confirmed one or more mutations in your dysferlin gene, please consult your physician. 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. In addition, the Jain Foundation (www.jain-foundation.org) also provides guidance to patients who are unsure about their diagnosis (contact Esther Hwang, Director of Patient Relations, at +1 425-882-1492 [US number] or ehwang@jain-foundation.org).

Why should I register?

- The more patient information collected, the better equipped researchers will be for finding therapies for this disease.
- You will receive information relevant to you or your condition – for example if researchers find better ways of caring for patients with a dysferlinopathy.
- You will be informed if you might be a suitable candidate for certain clinical trials or research studies.
- The data collected will reveal how many people worldwide have the same condition and provide relevant information to researchers interested in the best standards of care for your disorder.

Since dysferlinopathies are rare conditions, every single person counts!

What data are collected?

- Personal data, such as name, address and date of birth, so that patients can be identified and contacted by the Registry staff when appropriate.
- Medical data provided by the patient and relevant to their dysferlinopathy.
- After full consent by the patient, data related to their mutational and biological dysferlin tests will be provided by professionals who are involved in the diagnosis and follow-up of their condition.

Authorised third parties will be able to access unidentifiable medical data, and to use these data for research into dysferlinopathies or to help with the planning of and recruitment into scientific studies or clinical trials, but these third parties will not have access to patients' personal details, such as their name and contact information.

How do I register?

If you have a valid e-mail address, please register yourself or your child/dependent directly over the internet at www.dysferlinregistry.org. If you do not have internet access, paper forms are also available. All the registration forms can be downloaded from the Registry website, or you can contact the Registry to request all the forms by post. The Registry contact information is indicated on the inner fold of this leaflet.

To proceed with registration, you will be asked to read information documents, complete and validate/sign a consent form and fill in two online registration questionnaires. The Registry will then contact the doctor(s) you indicate in your questionnaires in order to obtain or confirm medical data required by the Registry. At any time, you can directly view or update your data online. You can contact the Registry at any time if you wish to withdraw your data.