JAIN FOUNDATION ORCHESTRATING A CURE
LGMD2B DYSFERLINOPATHY MIYOSHI

9725 Third Avenue NE, Suite 204 |Seattle, Washington 98115 | USA | Phone: 425-882-1492| Fax: 425-658-1703 | www.jain-foundation.org

Dear Clinician and Researcher:

You are receiving this letter because you have submitted sequence variants to the dysferlin database at the Leiden Muscular Dystrophy pages. We are writing to inform you about a foundation relevant to patients with mutations in the dysferlin gene and/or have a diagnosis of LGMD2B/R2/Miyoshi Myopathy which can be referenced as dysferlinopathies. The Jain Foundation (www.jain-foundation.org) was established to hasten development of a cure or therapy for dysferlinopathy. Our strategy is to:

1. Fund scientific research and the development of therapies/cures/drugs that target dysferlin deficiency (the cause of LGMD2B/R2/Miyoshi)
2. Curate the Dysferlin Registry and prepare the patient community for clinical trial recruitment
3. Advocate for inclusion of dysferlinopathy patients in clinical trials of existing therapies
4. Fund and direct a natural history study of dysferlinopathy (known as COS, the Clinical Outcome Study of Dysferlinopathy which began in 2013 and is an international study)
5. Provide support and access to diagnostic resources and information about dysferlinopathy

Because this is a rare disease with a prevalence of approximately 5-8/million, the symptoms, disease progression, and molecular analysis of each patient is very important to improving our understanding of the disease. In addition, through the registration of patients, we can determine the true incidence of LGMD2B/R2/Miyoshi and highlight its importance to scientists, clinicians, and drug/pharmaceutical companies worldwide. Furthermore, patient recruitment has become a limiting step for many clinical trials, especially trials that require knowledge of each patient's genetic mutations. Such mutation-targeted therapies are becoming much more common, and it is more and more difficult for patients who have not been fully diagnosed to the point of identifying mutations in both copies of their dysferlin gene to enroll in trials. Patient recruitment for clinical trials is particularly difficult in the case of rare diseases like LGMD2B/R2/Miyoshi.

The Jain Foundation is doing its best to address these issues for patients with dysferlinopathy, but cannot succeed without your help in reaching out to patients. Please point all of your patients with LGMD2B/R2/Miyoshi (with and without genetic confirmation) to the existence of the Jain Foundation: (https://www.jain-foundation.org/patient-physician-resources/patient-registration) Patients who register will have access to the following services:

- Information about ongoing or upcoming clinical trials relevant to their disease
- A source of knowledge about treatment options that are under development
- Access to diagnostic resources, specifically for unclear DYSF variants and dysferlin expression analysis
- General information about dysferlinopathy and patient to patient support

For additional information about the Jain Foundation, please feel free to contact Sarah Emmons at patients@jain-foundation.org or by phone in the U.S. area code (425) 882-1440. Thank you very much for your consideration and your efforts on behalf of these patients.