The LGMD2A/Calpainopathy Registry: A Patient-Powered Natural History Study and Trial Recruitment Tool

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Abstract

In September 2023, Coalition to Cure Calpain 3 launched the LGMD2A/Calpainopathy Registry. This global, patient-reported registry collects longitudinal data on individuals living with Calpainopathy (including limb-girdle muscular dystrophy type 2A/LGMDR1 and limb-girdle muscular dystrophy type 1I/LGMDD4).

The Registry utilizes the TREAT-NMD LGMD Core Dataset, the ACTIVLIM questionnaire, and the PROMIS Global-10 measure, as well as additional disease-specific items. Participants are encouraged to upload a digital copy of their genetic report. The registry curator reaches out to participants annually via email, requesting that they update their data on the platform. Participants can enter their own data, or, in the case of minors, Legally Authorized Representatives (LARs) can enter data on their behalf.

The LGMD2A/Calpainopathy Registry collects patient-entered data through a secure web-based application developed and maintained by the National Organization for Rare Disorders (NORD). It is compliant with U.S. Health Information Privacy Laws, FDA regulations on electronic records, and security requirements of General Data Protection Regulation (GDPR). There are no geographic restrictions to joining this global Registry. All patient-facing documents have been reviewed and approved by the North Star Review Board. A Steering Committee governs the Registry.

The Registry creates a platform to bring the Calpainopathy community together and collect patient data that is an essential requirement for policy makers, academic researchers, and pharmaceutical companies to advance treatments for this disease. Additionally, the study will help identify individuals with Calpainopathy who might be willing to take part in research studies or clinical trials. In the first four months after opening, 222 participants consented to join the Registry. Efforts around patient engagement and promotion of the Registry are ongoing to increase global participation. Data will be available for use with approval from the Steering Committee.

Objectives

- Collect longitudinal natural history data
- Support recruitment for clinical research studies

Surveys

Registration is patient-initiated at LGMD2A.IAMRARE.ORG.

Surveys utilize the TREAT-NMD LGMD Core Dataset[1], Patient-Reported Outcome Measures ACTIVLIM[2] and the PROMIS Global-10[3], and disease-specific items. Genetic testing reports may be uploaded in the Medical Reports survey.

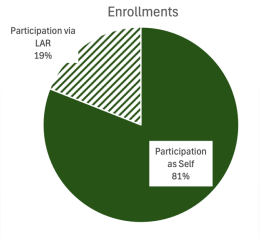
Surveys can be completed by adults or LARs. Updates are requested annually.

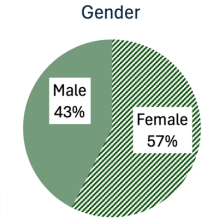
1. Profile (includes Global Unique Identifier items)
2. Demographics
3. Diagnosis
4. Family history
5. Symptoms and treatments
6. ACTIVLIM
7. PROMIS Global-10
8. Medical reports

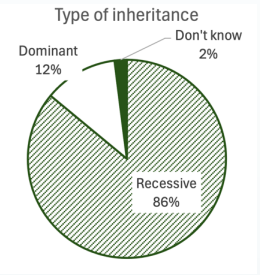
References

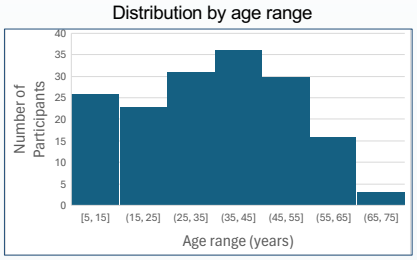
1. datasets.treat-nmd.org/lgmd/
2. ACTIVLIM adapted from Faculté de Médecine, Unité de Réadaptation et de Médecine Physique, UCL5375, Avenue Mounier 53, 1200 Bruxelles, Belgium. rehab-scales.org
3. healthmeasures.net/explore-measurement-systems/promis

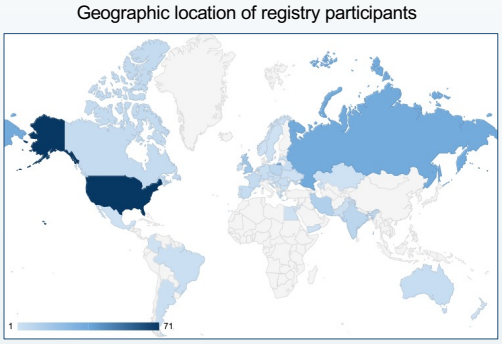
Results











Argentina	3
Australia	4
Austria	2
Belarus	4
Brazil	3
Bulgaria	1
Canada	5
Colombia	1
Croatia	2
Czech Republic (Czechia)	4
Egypt	1
France	5
Germany	3
Greece	2
Hungary	2
India	6
Iran, Islamic Republic of	12
Ireland	1
Jordan	1
Kazakhstan	1
Latvia	1
Mexico	1
Netherlands	2
New Zealand	2
Norway	1
Pakistan	2
Poland	6
Portugal	2
Romania	3
Russian Federation	24
Serbia	2
Slovakia	1
Spain	4
Sweden	1
Switzerland	1
Ukraine	1
United Kingdom	17
United States	71
Yemen	1

Conclusions

- The LGMD2A/Calpainopathy Registry is a valuable tool to collect patient-reported information.
- The Registry collects data from a large and diverse cohort, representing a range of all ages and geographic locations. This facilitates the participation of individuals not typically included in clinical studies.
- Utilization of the TREAT-NMD Core Dataset and validated Patient Reported Outcome measures will enable the harmonization of Registry data with data collected from other sources.

Future Directions

- Identify recruitment strategies to expand participation
- Curate genetic testing reports
- Support additional languages (French and Spanish translations in progress)
- Analyze and publish baseline and longitudinal data
- Facilitate clinical trial recruitment
- Support requests for de-identified patient data

Contact Registry@CureCalpain3.org with inquiries or to request print materials.

LGMD2A.IAMRARE.ORG