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

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Introduction

In September 2023, Coalition to Cure Calpain 3 launched the LGMD2A/Calpainopathy Registry. This global, patient-reported registry has two main goals:

1. **Collects data over time from individuals living with Calpainopathy (including limb-girdle muscular dystrophy type 2A/LGMDR1 and limb-girdle muscular dystrophy type 1I/LGMDD4)**
2. **Supports recruitment for clinical research studies.**

The Registry collects information about motor function, activity, and quality of life. 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 can enter data on their behalf.

Methods

Registration is patient-initiated at LGMD2A.IAMRARE.ORG, a secure web-based application developed and maintained by the National Organization for Rare Disorders. It is compliant with U.S. Health Information Privacy Laws, FDA regulations on electronic records, and security requirements of General Data Protection Regulation (GDPR). The Registry is available globally. All patient-facing documents have been approved by the North Star Review Board. A Steering Committee governs the Registry.

Recruitment Support

The Registry has been utilized to assist in participant recruitment for three studies:

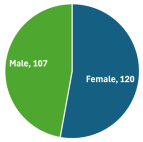
1. An industry-sponsored natural history study
2. An academic-sponsored natural history study
3. A study to apply genetic sequencing on samples from undiagnosed or partially diagnosed individuals with suspected LGMD

Recruitment materials were shared with Registry Participants residing in eligible countries who opted-in to receive news about studies.

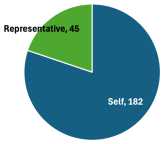
Results

Data entered by 227 Participants
Average age: 35 ± 16 years

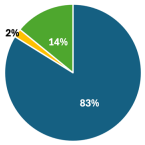
Sex



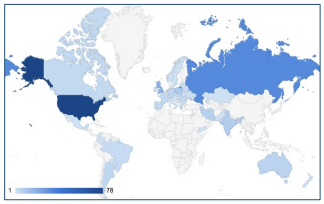
Type of Registration



Participant Status

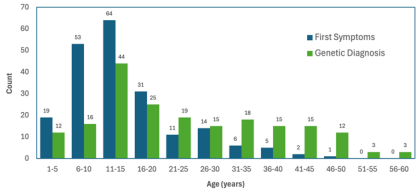


41 Countries Represented



United States	78	Greece	2
Russian Federation	33	New Zealand	2
United Kingdom	18	Ireland	1
Poland	10	Bulgaria	1
India	8	Mexico	1
Iran, Islamic Republic of	7	Slovakia	1
Australia	6	Hungary	1
Germany	5	Italy	1
Argentina	5	Austria	1
France	4	Pakistan	1
Spain	4	Sweden	1
Canada	4	Paraguay	1
Netherlands	3	Norway	1
Romania	3	Switzerland	1
Brazil	3	Colombia	1
Belarus	3	Lithuania	1
Czech Republic (Czechia)	3	Jordan	1
Croatia	2	Yemen	1
Serbia	2	Kazakhstan	1
Portugal	2	Latvia	1
Ukraine	2		

Age Symptoms First Noticed and Age at Diagnosis of Calpainopathy



Conclusions

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. In the first twelve months after opening, data was entered by over 200 participants. Efforts around patient engagement and promotion of the Registry are ongoing to increase global participation.

Data collected through the Registry describe people who have Calpainopathy to give researchers a better understanding of the condition, the different way it affects people, and how it changes over a person's lifetime. This information is helpful to develop management guidelines to improve the care and quality of life for people living with Calpainopathy. Data will be available for use with approval from the Steering Committee.

Additionally, the study helps identify individuals with Calpainopathy who might be willing to take part in research studies or clinical trials. To-date, it has been utilized to assist in participant recruitment for three clinical studies.

Future Directions

- Identify strategies to expand participation
- 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