LGMD CORE DATASET PROJECT

Objectives survey answers-Patient representatives
Jennifer Levy-Coalition to Cure Calpain 3
List of participants

- Global FKRP Registry
- Dysferline registry-Jain Foundation
- AFM-Téléthon
- LGMD2A/R1 Global Registry-Coalition to cure Calpain 3
- LGMD2L Foundation
- LGMD1D DNAJB6 / MYOSYND™ : International Autosomal Dominant and X Linked Muscular Dystrophy Registry-LGMD1D DNAJB6 Foundation
- LGMD Registry-UILDM - Unione Italiana Lotta alla Distrofia Muscolare Italy
- Spanish Federation of Neuromuscular Diseases (ASEM Federation)
Survey questions

• Please rank the possible objectives of a registry in order of **importance** to you

• Please, try now to select **only three objectives** which you consider to be the most essential for a LGMD registry

• With a view to **feasibility** planning, please rate each objective according to **how easy** it will be to deliver?

• With a view to **feasibility** planning, please indicate your view on what a realistic **timeframe** might be for implementation of each aim?

**Objectives**

• **Clinical characterization of the diseases** *(description of the disease characteristics such as onset of the disease and phenotype)*

• **Longitudinal data collection/Natural history of the diseases** *(collection of follow-up data describing the evolution of the disease over time)*

• **Assessment of Cost-effectiveness of treatments** *(understanding of monetary costs of disease-modifying drugs relative to their effectiveness in achieving a predefined purpose.)*

• **Health care and social planning for patients** *(assessment of patient healthcare and social needs, existing resources and their applicability to patients.)*

• **Recruitment for clinical trials and feasibility of clinical trials**

• **Conducting clinical research** *(use of the registry to answer ad-hoc research questions)*

• **Support Biobanks and basic research** *(genetic, molecular, psychological basis of the disease)*

• **Post-marketing surveillance**

• **Collection of Patient reported outcome measures and quality of life** *(monitoring of the safety and efficacy of treatment following ethical approval)*

• **Developing a community of patients, families, and clinicians**
Please rank the possible objectives of a registry in order of **importance** to you.
Please, try now to select **ONLY THREE objectives** which you consider to be the most essential for a LGMD registry.
With a view to **feasibility** planning, please rate each objective according to **how easy** it will be to deliver?
With a view to **feasibility** planning, please indicate your view on what a **realistic timeframe** might be for implementation of each objective?
## Summary

<table>
<thead>
<tr>
<th>Objective</th>
<th>Main objectives</th>
<th>Feasibility</th>
<th>Feasibility</th>
</tr>
</thead>
<tbody>
<tr>
<td>Recruitment for clinical trials</td>
<td>8 votes</td>
<td>Easy (6)</td>
<td>Long term (8)</td>
</tr>
<tr>
<td>Conducting clinical research</td>
<td>3 votes</td>
<td>Easy (5)</td>
<td>Medium-short term (5)</td>
</tr>
<tr>
<td>Clinical characterization of the disease</td>
<td>3 votes</td>
<td>Easy (3)/Difficult (3)</td>
<td>Long term (5)</td>
</tr>
<tr>
<td>Longitudinal data collection</td>
<td>3 votes</td>
<td>Easy (3)/Difficult (3)</td>
<td>Medium-short term (4)</td>
</tr>
</tbody>
</table>
Conclusions

• All participants agreed on the main objective as the recruitment of patient in clinical trials

• The criteria used to rank the objectives for importance were:
  ▪ scientific and clinical aspects (increase our knowledge of the disease)
  ▪ Address immediate needs
  ▪ Create a community and keep patients informed

• It is important to collaborate with clinicians to understand what is most important to collect to address the gaps in our understanding of the diseases

• It is easy to collect patient reported data but more difficult to have clinicians' input
LGMD CORE DATASET PROJECT

Objectives survey answers-Clinicians and registry curator
Meredith James-John Walton Muscular Center
List of participants

- Belgian Neuromuscular diseases registry (Registry curator)
- Canadian Neuromuscular diseases registry (Registry curator)
- Egyptian Neuromuscular registry (Registry curator)
- John Walton Muscular Center (Physician/ Physiotherapist-Advisory Board)
- Kennedy Krieger Institute (Physician)
- Friedrich-Baur-Institute, Dept. of Neurology, Klinikum der Universität Munich, Germany (Physician-Advisory Board)
- Neuromuscular Disease Natural History Study (Nationwide Children Hospital) (Registry curator)
- New Zealand Neuromuscular Disease Registry (Registry curator)
- Hospital Pitie-Salpetriere (Physician-Advisory Board)
- Spanish Neuromuscular Diseases Registry (Registry curator)
- Australian Neuromuscular Disease Registry (Registry curator)
- Copenhagen Neuromuscular Center (Physician-Advisory Board)
- University Medical Center Ljubljana (Physician)
- UOD Neurologia-Malattie Neuromuscolari e Rare Neurologia, Fondazione I.R.C.C.S. Ca’ Granda Ospedale Maggiore Policlinico.Università degli Studi di Milano (Physician)
Please rank the possible objectives of a registry in order of importance to you.

Confirmation of the diagnosis/ Genetic diagnosis of patients
Please, try now to select **ONLY THREE objectives** which you consider to be the most essential for a LGMD registry.

### Main objectives

<table>
<thead>
<tr>
<th>Objective</th>
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<td>Developing a community of patients,</td>
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<tr>
<td>Collection of Patient reported outcome</td>
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<tr>
<td>Post-marketing surveillance</td>
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<tr>
<td>Support Biobanks and basic research</td>
<td></td>
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<tr>
<td>Conducting clinical research</td>
<td></td>
</tr>
<tr>
<td>Recruitment for clinical trials and feasibility</td>
<td></td>
</tr>
<tr>
<td>Health care and social planning for patients</td>
<td></td>
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<tr>
<td>Assessment of Cost-effectiveness of</td>
<td></td>
</tr>
<tr>
<td>Longitudinal data collection/Natural</td>
<td></td>
</tr>
<tr>
<td>Clinical characterization of the diseases</td>
<td></td>
</tr>
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With a view to **feasibility** planning, please rate each objective according to **how easy** it will be to deliver?
With a view to feasibility planning, please indicate your view on what a realistic **timeframe** might be for implementation of each objective?

<table>
<thead>
<tr>
<th>Objective</th>
<th>Long term</th>
<th>Medium-short term</th>
<th>NA</th>
</tr>
</thead>
<tbody>
<tr>
<td>Developing a community of patients, ...</td>
<td></td>
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*Note: The chart shows varying timeframes for each objective with different color codes.*
Summary

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<th>Feasibility</th>
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<td>Recruitment for clinical trials</td>
<td>11 votes</td>
<td>Easy (9)</td>
<td>Medium-short term (13)</td>
</tr>
<tr>
<td>Longitudinal data collection</td>
<td>9 votes</td>
<td>Difficult (8)/ easy (5)</td>
<td>Long term (9)</td>
</tr>
<tr>
<td>Clinical characterization of the disease</td>
<td>7 votes</td>
<td>Easy (10)</td>
<td>Medium-short term (13)</td>
</tr>
<tr>
<td>Conducting clinical research</td>
<td>5 votes</td>
<td>Easy (7)/ difficult (4)/ very easy (3)</td>
<td>Long term (9)</td>
</tr>
<tr>
<td>Collection of PROM</td>
<td>2 votes</td>
<td>Easy (12)</td>
<td>Medium-short term (12)</td>
</tr>
</tbody>
</table>
Conclusions

• There are many unanswered questions about LGMDs. The registry can help us learn more about these diseases and be ready for future treatments.

• The design of a clinical data set is challenging and the feasibility of collecting this data may be different at each site.

• Important to consider the different characteristic of the registries in order to assess the feasibility of the collection of core dataset.
Conclusions

• The core dataset must be flexible to adapt to the evolving field, the types of LGMD and the different realities between registries. MINIMUM CORE DATASET FOR LGMD (expanded dataset in the future)

• Important to differentiate what is important to collect from what is feasible to collect.

• The collection of longitudinal data (natural history of the disease) although it is very important is difficult to achieve.
LGMD CORE DATASET PROJECT

Objectives survey answers-Pharmaceutical companies
James Richardson-Sarepta
List of participants

- Affinia
- Askbio
- Catabasis
- Sarepta
- Edgewise
Please rank the possible objectives of a registry in order of importance to you.

- Developing a community of patients...
- Collection of Patient reported outcome...
- Post-marketing surveillance
- Support Biobanks and basic research
- Conducting clinical research
- Recruitment for clinical trials and feasibility...
- Health care and social planning for patients
- Assessment of Cost-effectiveness of...
- Longitudinal data collection/Natural history...
- Clinical characterization of the diseases

**emphasis importance in collection of genotypic data**
Please, try now to select **ONLY THREE objectives** which you consider to be the most essential for a LGMD registry.
With a view to feasibility planning, please rate each objective according to how easy it will be to deliver?
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<td>5 votes</td>
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<td>Long term (3)</td>
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<tr>
<td>Recruitment for clinical trial</td>
<td>3 votes</td>
<td>Easy (3)/difficult (2)</td>
<td>Short-Medium term (5)</td>
</tr>
<tr>
<td>Collection of PROM</td>
<td>2 votes</td>
<td>Difficult (4)</td>
<td>Short medium term (3)</td>
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<td>Clinical characterization of the disease</td>
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Conclusions

• It is a priority to have robust and reliable natural history data/longitudinal data of the diseases to support multiple aspects of the clinical development of treatments, including its possible use as a control group for clinical trials.

• Registries could be used to identify the most meaningful outcome measures for patients. If they are identified with patient involvement, are collected in a standardised and reliable way, they are more likely to be accepted by regulators.

• Clinical characterization and longitudinal data collection should include clinical, genetical, and molecular characterization of the disease.
Conclusions

- Linking registries and biobank samples could play an important role in drug development facilitating the identification of biomarkers of disease progression or drug effect.
- Planning the collection of biobank samples in conjunction with registry data needs to be considered in advanced but can complement clinical data.
- Combined patient and clinician reported registries are more likely to collect robust data which can be acceptable from a regulatory point of view. Joint efforts to achieve the same goal.
- Different stakeholders have different perspectives and motivations, and it is interesting to understand the perspective of others, their needs and their background.