Establishing the REaDY LGMD Registry: A Czech National Database for Limb Girdle Muscular Dystrophies

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Introduction

Limb Girdle Muscular Dystrophies (LGMD) are a group of rare, genetically and clinically heterogeneous neuromuscular diseases. Due to their **orphan status**, each patient represents a valuable source of information for improving care and advancing research.
As **gene-targeted therapies approach** clinical use, healthcare systems must be **ready to identify** eligible **patients** and **monitor treatment** outcomes. This requires high-quality, structured, and standardized longitudinal data.
The REaDY LGMD Registry was established to ensure a **unified standard of care**, enable **clinical trial readiness**, and integrate the Czech Republic into **international research efforts.**

Methods

The **REaDY LGMD Registry** was launched in June 2020 as a **national, clinician-driven database** to collect data on genetically confirmed LGMD cases. It is **bilingual** (Czech/English), **ethically approved**, and operated under the Czech Neurological Society and the Czech Society for Paediatric Neurology. **Seven neuromuscular centres** contribute data.

Collected data domains include:
- **Demographics**
- **Genetic diagnosis** (certified labs)
- **Functional status**
- **Disease progression**
- **Diagnostic timelines**
- **Patient-reported outcomes**

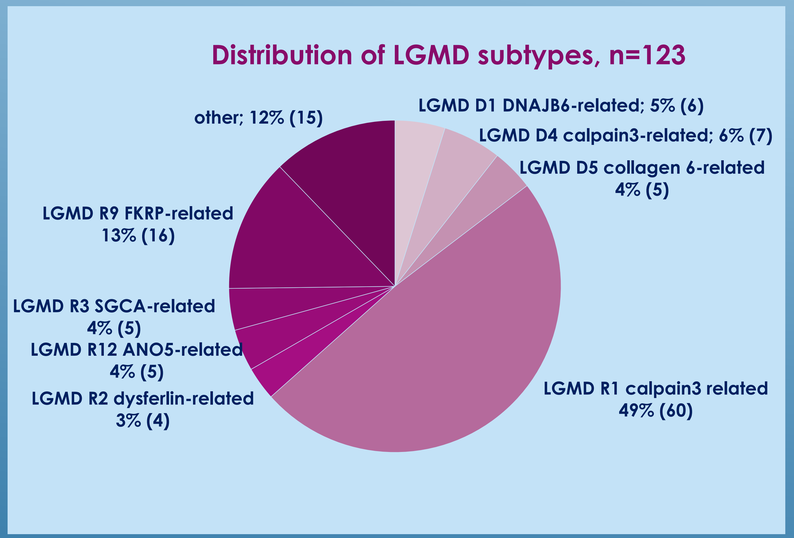
Technical and analytical oversight is provided by the **Institute of Biostatistics and Analyses (IBA s.r.o.),** which also serves as the data processor. Data quality is reviewed quarterly, and annual curator workshops ensure continuous improvement and alignment.

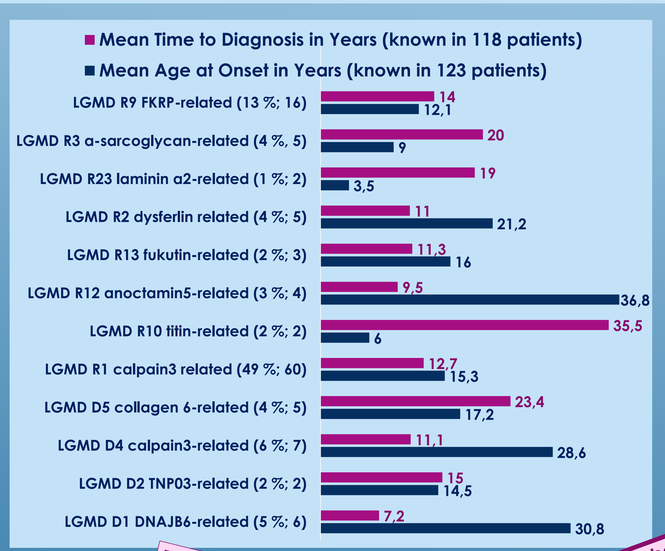
The registry's **dataset structure** follows the standardized documentation model used in **TREAT-NMD**. Since 2022, the Czech Republic has been a **core membe**r of this network, enabling seamless inclusion of registry patients into international studies.

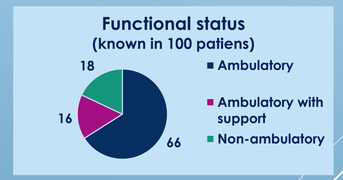
Patients have **secure access** to their own records and can independently complete the SF-36 quality-of-life survey. Initial enrollment exam is followed by mandatory follow-ups at least every two years for validity.

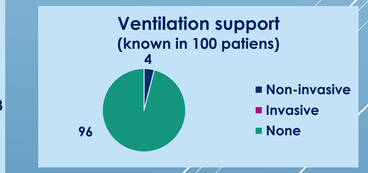
Results

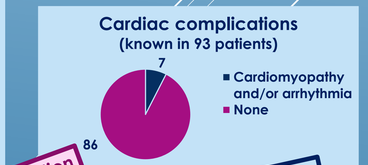
As of May 5, 2025, the REaDY LGMD registry included data on **123 patients** with **genetically confirmed** LGMD diagnosis, consisting of **54 males** (43.9 %) and **69 females** (56.1 %) from **7 neuromuscular centers** across the Czech Republic. **The most common LGMD subtypes were:** LGMD R1 calpain3-related: 60 patients (48.8%), LGMD R9 FKRP-related: 16 patients (13.0 %), LGMD D4 calpain3-related: 7 patients (5.7 %),
LGMD D1 DNAJB6-related: 6 patients (4.9 %), LGMD D5 collagen 6-related, LGMD R2 dysferlin-related and LGMD R3 a-sarcoglycan-related: each 5 patients (4.1 %). **The median age at symptom onset** was **12 years** (range 1–56), and the **median age at diagnosis** was **28 years** (range 1–70). There was **no statistically significant difference** in the time from symptom onset to diagnosis between LGMD subtypes ($p = 0.233$, Kruskal–Wallis test**). Functional status** data were available for 100 of the 123 patients. Among them, 66 (66.0%) were ambulatory, 16 (16.0%) ambulatory with support, and 18 (18.0%) non-ambulatory.
Cardiovascular complication data were available for 93 patients, among whom 7 (7.5%) had cardiomyopathy and/or arrhythmia. **Ventilation support data** were available for 100 patients; 4 patients (4.0%) had used or were currently using non-invasive ventilatory support. No patients required invasive ventilation.











Conclusion

The REaDY LGMD Registry represents the **first structured national database of genetically confirmed LGMD patients in the Czech Republic**. Initial data show a predominance of CAPN3-related LGMD and highlight substantial diagnostic delay, despite most patients remaining ambulant at the time of registration.
The registry serves as a foundation for **improving national standards of care**, ensuring **readiness for clinical trials**, and supporting **international collaborations.**
With gene therapies increasingly targeting paediatric populations, we emphasize the importance of enrolling children early to enable timely access to trials and emerging therapies.
Future goals include expanding **patient recruitment**, enhancing data completeness through regular **follow-ups**, and contributing to global analyses via the TREAT-NMD network.

For current registry data, scan here.

Collaboration welcome!

The REaDY LGMD Registry supports structured and longitudinal data collection, including genetic diagnosis, functional assessments, and ancillary diagnostics (muscle biopsy, cardiac imaging, MRI, etc.).

We welcome collaborative projects focused on data analysis, biomarker validation, or clinical study planning.

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Conflicts of interest
The REaDY LGMD Registry is supported by institutional funding and unrestricted research grants from multiple industry partners. No commercial sponsor had any influence on data collection, analysis, or interpretation.