# Patient Registries and their Importance in Translational and Clinical Research

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Neuromuscular diseases (NMD) are a heterogeneous group of genetic conditions characterized by progressive muscle degeneration and weakness. In Europe, all NMD have a prevalence of ≤5 per 10,000 and are therefore considered rare. Patient registries are research databases containing demographic, genetic and clinical information about individuals affected by a condition or genetic mutation, and can facilitate translational research in rare neuromuscular diseases by:

- Rapidly locating participants for **clinical trials** and **research studies**.
- Providing **natural history** and **epidemiological** data.
- Answering specific **research** questions from **academics, healthcare professionals** and **industry**.
- Collecting **real world data** to inform regulatory pathways and monitor the safety and efficacy of new treatments.
- Informing the understanding and development of standards of care.
- Linking the research community to the patients by offering a two-way flow of disease-specific information.

**Patient registry data can be made available for academic or commercially funded research—please get in touch with the registries team to discuss how we can support your project.**

Since 2008 the JWMDRC patient registries have been involved in **over 50** scientific publications, and **over 75** research enquiries including, but not limited to:

⇒ Clinical trial recruitment
⇒ Dissemination of clinical and academic research surveys
⇒ Provision of de-identified data to researchers for analysis and publication.

Examples of registry enquiries include:

- Recruitment onto phase I, II and III clinical trials in multiple disease areas
- Obtaining patient feedback on a clinical trial protocol
- Recruitment onto non-interventional research studies on topics such as wearable activity monitors, accessibility issues in the built environment, and pregnancy in NMD.

Contact the JWMDRC registries team:
registries@ncl.ac.uk
0191 241 8640

The John Walton Muscular Dystrophy Research Centre (JWMDRC) manages seven national and international patient registries comprising **over 4,300** patients. All registries are affiliated with the global neuromuscular network TREAT-NMD and conform to their internationally standardised core datasets, and the three national registries contribute data to the TREAT-NMD Global Registries Network (TGDOC).

## UK National Patient Registries*

** UK registries may also accept registrations from international patients if there is no suitable registry in their country; they are not included in the numbers below*

**UK SMA Patient Registry**

- SMA Type 0
- SMA Type 1
- SMA Type 2
- SMA Type 3a/3b
- SMA Type 4
- SMA Type undetermined

Now collecting PROMs

636 patients

www.sma-registry.org.uk

**UK FSHD Patient Registry**

- FSHD Type 1
- FSHD Type 2
- FSHD type undetermined

906 patients

www.fshd-registry.org/uk

**UK Myotonic Dystrophy Patient Registry**

- Myotonic Dystrophy Type 1
- Myotonic Dystrophy Type 2
- Congenital Myotonic Dystrophy
- Mutation carrier for DM1 without symptoms
- Myotonic Dystrophy Type undetermined

839 patients

www.dm-registry.org/uk

Plans are underway to further extend the registries' research support capabilities through data linkage to the Newcastle Research Biobank for Rare and Neuromuscular Diseases and other initiatives. Work to establish the registries as a Newcastle University Core Research Facility will improve the sustainability and visibility of the registries and increase income generation potential. Future plans also include the development of the registries as Clinical Trial Recruitment Hubs, to support recruitment and equity of access in neuromuscular research, and the use of registry data in support of regulatory post-marketing surveillance of new therapies.

Patient numbers correct as of 24/03/2023

## International Patient Registries

**Global FKRP Registry**

- Limb-girdle muscular dystrophy type R9 or 2I (LGMDR9 or LGMD2I)
- Congenital Muscular Dystrophy Type 1C (MDC1C)
- Muscle Eye Brain Disease
- Walker-Warburg Syndrome

916 patients representing 50 countries

www.fkrp-registry.org

**The Myotubular and Centronuclear Myopathy Patient Registry**

- X-linked myotubular myopathy (XLMTM)
- Centronuclear myopathy (CNM)
- Female carriers of XLMTM

*Available in 10 languages*

448 patients representing 53 countries

www.mtmcnmregistry.org

**Global Registry for COL6-related dystrophies**

- Ullrich congenital muscular dystrophy (UCMD)
- Bethlem myopathy
- Bethlem/Ullrich intermediate

196 patients representing 39 countries

www.collagen6.org

**GNE Myopathy International Patient Registry**

- GNE myopathy
- Hereditary inclusion body myopathy (HIBM)
- Nonaka myopathy
- Distal myopathy with rimmed vacuoles (DMRV)

424 patients representing 37 countries

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Muscular Dystrophy UK — Fighting muscle-wasting conditions
astellas GENE THERAPIES
CURELGMD2i Limb Girdle Muscular Dystrophy
Myotonic Dystrophy Support Group
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LGMD2iFund Limb Girdle Muscular Dystrophy 2I Research Fund
Biogen
myotubular trust FINDING STRENGTH
Cure DM Research ♥ Awareness ♥ Support Cure Myotonic Dystrophy UK Charity