

# WHY JOIN A PATIENT REGISTRY?

Patient registries are databases containing information about individuals who are affected by a specific condition or genetic mutation. In **rare diseases**, they play an important role in the **development of new therapies**. The neuromuscular registries listed below are coordinated from **the John Walton Muscular Dystrophy Research Centre**.

## Registries can:

- Identify participants for **clinical trials**.
- Help develop **care standards**, to help improve the care people receive.
- Support specific **research** questions for **doctors and scientists**.
- Contribute to the **natural history** of a rare disease.
- Provide a **link to the research community** enabling people to receive information directly relevant to their condition (for example, newsletters or updates on standards of care).

## UK National Patient Registries



**UK SMA**  
PATIENT REGISTRY

[www.sma-registry.org.uk](http://www.sma-registry.org.uk)



**UK FSHD**  
Patient Registry

[www.fshd-registry.org/uk](http://www.fshd-registry.org/uk)



**UK Myotonic Dystrophy**  
Patient Registry

[www.dm-registry.org/uk](http://www.dm-registry.org/uk)

## International Patient Registries

**GLOBAL FKR**  
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



[www.fkrp-registry.org](http://www.fkrp-registry.org)

The Myotubular and Centronuclear Myopathy Patient Registry

- X-linked myotubular myopathy (MTM)
- Centronuclear myopathy (CNM)



[www.mtmcnmregistry.org](http://www.mtmcnmregistry.org)

Global Registry for COL6-related dystrophies

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



[www.collagen6.org](http://www.collagen6.org)

**GNE** Myopathy International Patient Registry

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



[www.gne-registry.org](http://www.gne-registry.org)

## Examples of how patient registries have been used successfully:

- The **UK DM Registry** has previously been used to support the recruitment onto a phase II clinical trial of tideglusib in teenagers and adults with congenital and childhood-onset DM. The registry has also been used to support a falls and fall-associated fractures survey in patients with DM1.
- The **UK FSHD Registry** was used to help a pharmaceutical company gain patient insight into their upcoming clinical trial protocol. This was captured via a survey sent out through the registry.
- The **UK SMA Registry** was used to distribute an EU wide survey regarding patient quality of life and pain.
- The **Global FKR Registry** was used to support participant recruitment onto a phase III clinical trial of deflazacort in adults with LGMD2I.
- The **GNE Myopathy Registry** was used to support participant recruitment onto a phase III clinical trial of aceneuramic acid in adults with GNE myopathy.
- Many of the registries have collectively supported participant recruitment onto a research project investigating activity monitoring in patients with neuromuscular conditions.

For any questions please contact: [registries@ncl.ac.uk](mailto:registries@ncl.ac.uk)