The UK Myotonic Dystrophy Patient Registry - Empowering Clinical Research and Patient Voice with an Effective Translational Research Tool

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Background

What is the UK Myotonic Dystrophy Patient Registry?

The UK Myotonic Dystrophy (DM) Patient Registry is a patient-initiated, clinically supported national platform, collecting data from myotonic dystrophy patients. Established in May 2012, the registry is coordinated by the **John Walton Muscular Dystrophy Research Centre (JWMDRC)** at Newcastle University.

What are the registry aims?

- Support academic and clinical research
- Improve understanding and characterisation of DM
- Identify eligible patients for clinical trials
- Disseminate information regarding studies and research advancements to patients

Methodology

The registry captures longitudinal, self-reported data via a secure online portal. Patients are encouraged to update their questionnaires annually and neuromuscular specialists are invited to provide patients' clinical and genetic information. The registry is a Core Member of the **TREAT-NMD Global Registries Network** for Myotonic Dystrophy, collecting the *standardised core dataset* and contributing to global data enquiries.

The registry was transitioned to a new software platform in April 2025; however, the aggregate **patient reported data** presented reflects active, UK-based patient entries up to **March 2025**. Response rates are shown by denominator in figure titles.

Overcoming Barriers to Participation

The UK Myotonic Dystrophy Patient Registry is continuing to work on increasing patient engagement by:

- Transitioning to a custom registry platform designed for better patient experience, with improved usability and built-in data quality checks
- Promoting the registry at patient engagement days
- Inviting more doctors to participate in registry
- Allowing patients to fill out medical questionnaires with doctor's letter as verification

Conclusion

The registry serves as a **valuable research tool**, functioning as a large national DM registry that plays a crucial role in supporting data collection. It enables **efficient and standardised data gathering** from both **patients and clinicians**, ensuring comprehensive insights.

Additionally, the registry facilitates various research enquiries, as its data is aligned with **FAIR data standards** and global TREAT-NMD dataset requirements, making it widely **accessible and interoperable**.

Beyond data collection, the registry actively **promotes patient engagement** by providing a platform for individuals to contribute their experiences, stay informed about ongoing research, and participate in studies that could impact future treatments. It also serves as a channel for **disseminating information** about relevant events, conferences, and advancements in the field, ensuring that both **patients and healthcare professionals remain informed and connected.**

Results

There are 903 active patients in the registry

- 427 male patients (47.3%) and 476 female patients (52.7%)
- Average age of participants is 46.8 ± 17.6 years old (range 0-87)
- Confirmation of genetic diagnosis received from 364 patients (40.3%)

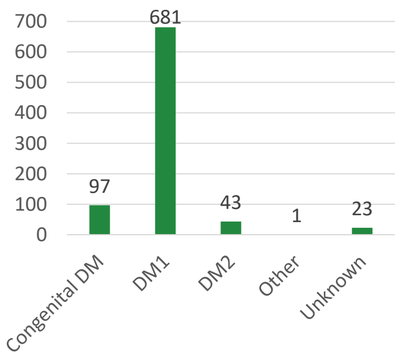

Figure 1: Clinical diagnosis of registry participants *n=845*

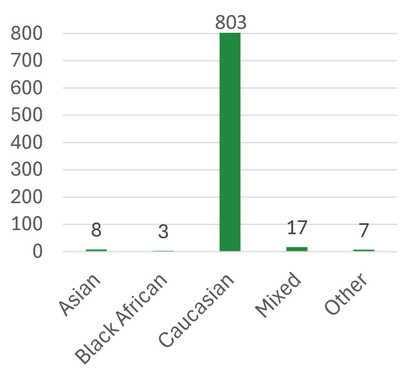

Figure 2: Ethnicity of registry participants *n=838*

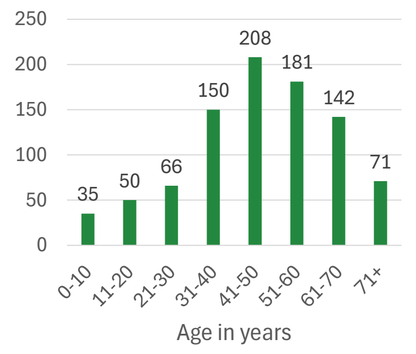

Figure 3: Age distribution of registry participants *n=903*

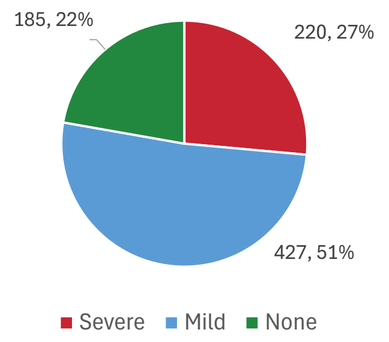

Figure 4: Fatigue experienced by registry participants *n=832*

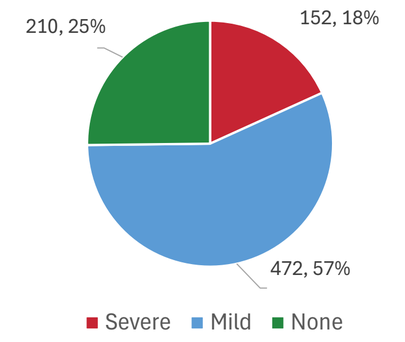

Figure 5: Myotonia experienced by registry participants *n=834*

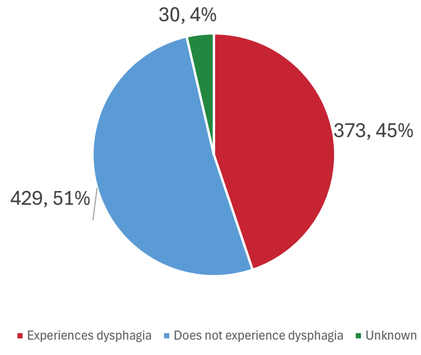

Figure 6: Dysphagia experienced by registry participants *n=832*

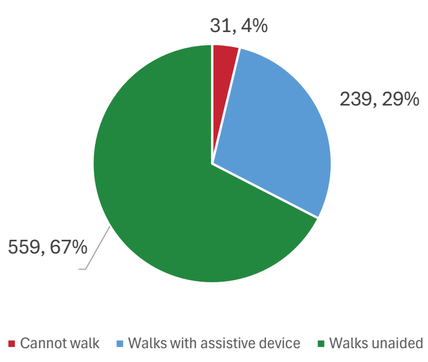

Figure 7: Current best motor function of registry participants *n=829*

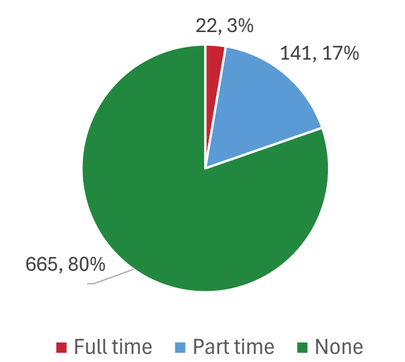

Figure 8: Wheelchair usage of registry participants *n=828*

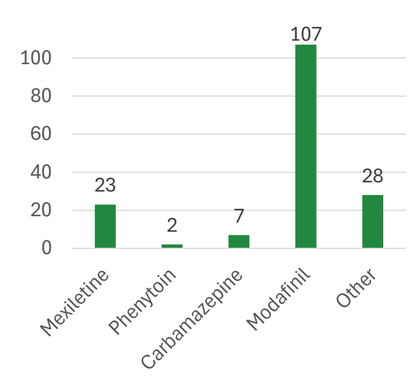

Figure 9: Medication used by registry participants *n=832*

The majority of patients in the registry have been diagnosed with DM1, while DM2—typically more prevalent in Eastern Europe—is observed in only a small subset. The registry primarily consists of adults, as myotonic dystrophy is less common in children. Fatigue is the most frequently reported symptom, with 78% of respondents experiencing mild to severe fatigue. Accordingly, fatigue management medications are the most commonly used medicine within this cohort. Additionally, myotonia is reported by 75% of respondents, whereas dysphagia is observed in 45% of cases.

The registry continues to support enquiries from academic and industry researchers. A few recent collaborations we have supported are:

- Clinical trial recruitment support
- International collaboration to improve knowledge on myotonic dystrophy type 2. *Journal of Neuromuscular Diseases*
- Increased frequency of repeat expansion mutations across different populations. *Nature Medicine.*

We are grateful to our research participants, steering committee members, patient advisors, and wider study team.

This registry is funded by:

To get in touch with the team, contact us at myotonicdystrophyregistry@ncl.ac.uk or visit www.dm-registry.org.uk