The UK Myotonic Dystrophy Patient Registry: Data Collection and Collaboration

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Background

- The UK Myotonic Dystrophy Patient Registry is a patient self-enrolling online database, established in May 2012 to collect clinical and genetic information about myotonic dystrophy type 1 (DM1) and myotonic dystrophy type 2 (DM2).
- Supported by Muscular Dystrophy UK (MDUK), Cure Myotonic Dystrophy UK Charity (Cure DM) and the Myotonic Dystrophy Support Group (MDSG), and coordinated by the John Walton Muscular Dystrophy Research Centre at Newcastle University.
- The registry's primary aim is to facilitate and accelerate clinical research in DM1 and DM2.
- The registry also aims to better characterise and understand DM, and disseminate information relating to upcoming academic, clinical and and non-clinical studies in DM.

Method

- The registry is used to capture **longitudinal**, **self-reported** data through an online portal available to patients and specialist clinicians.
- **Patient reported outcomes** are entered into a secure portal, combined with clinician verified genetic details. Patients are reminded to update their information annually, and all registrations are confirmed by the registry curator.
- Data collected within the registry includes all mandatory and highly encouraged items in the TREAT-NMD Core Dataset for Myotonic Dystrophy (2009), including both **patient reported**, and **doctor reported items** such as genetic confirmation.
- The registry is also now able to receive **genetic reports** directly from patients via a secure file upload link.
- The registry can support researchers and industry with a wide range of projects, including creation of **de-identified patient data reports** for use in feasibility studies, dissemination of research surveys, and trial recruitment support.

Demographics

Patient Numbers

As of Nov 2022, there are **840** active UK based participants enrolled on the UK Myotonic Dystrophy Patient Registry.

This includes 410 male and 430 female participants (a 49/51%) split).

The average age is 46.3 years (Male = 46.4 years, Female = 46.2 years).

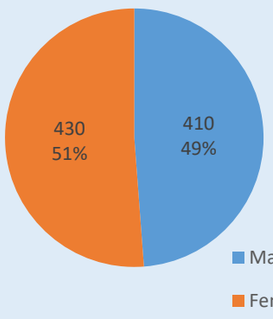

Figure 1. Self-reported patient sex at registration

Clinical / Genetic Diagnosis

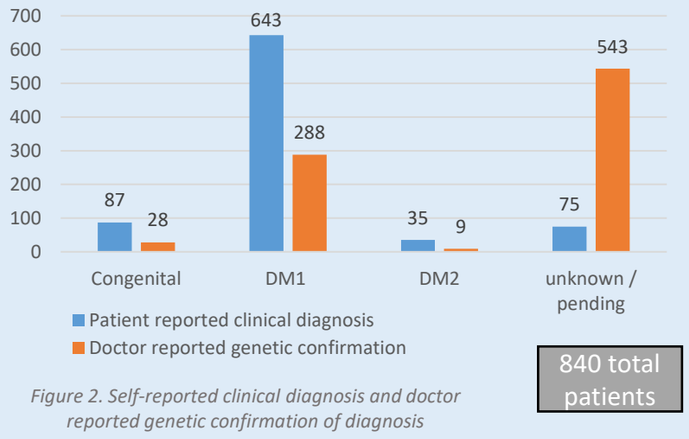

Figure 2. Self-reported clinical diagnosis and doctor reported genetic confirmation of diagnosis

840 total patients

The most common self-reported **clinical diagnosis** is DM1 (76.5%) followed by Congenital DM1 (10.4%), then DM2 (4.2%). Clinical diagnosis has not yet been reported for 8.9% of patients, who are being followed up. **Genetic confirmation** has so far been provided for 322 **(39%)** of all patients; extensive communication with patients and clinicians is underway to increase this, and the recent launch of our secure file upload site should lead to more reports being shared directly with the registry.

Family History

81% percent of patients reported a **positive family history** of DM, with 12% reporting no family history, and 7% unknown.

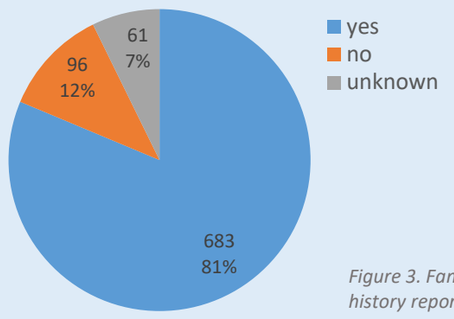

Figure 3. Family history reported

Pregnancies

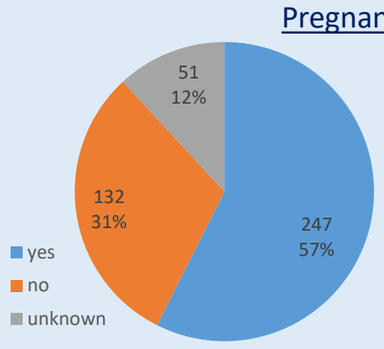

Figure 4. Self-reported pregnancies

57% percent of female patients reported at least one **pregnancy**, 31% reported no pregnancies, and 12% did not answer.

Longitudinal data and completeness

325 registered patients **(39%)** have updated their registry record over the last 24 months, and a further 221 **(26%)** have updated in at least the last 5 years.

Longitudinal data is available for at least 455 **(54%)** of patients to date.

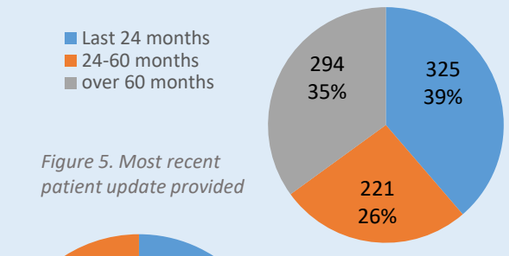

Figure 5. Most recent patient update provided

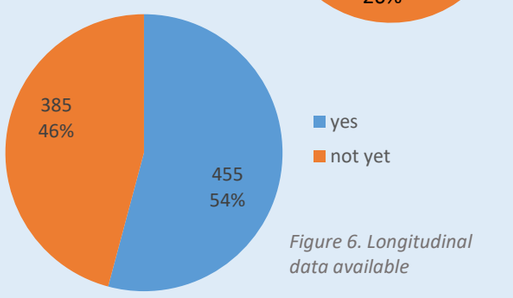

Figure 6. Longitudinal data available

Registry Use and Engagement with TREAT-NMD

To date the UK DM Patient Registry has supported at least **31** enquiries from industry, academics, clinicians and patient organisations. Most registry enquiries have involved online survey distribution (48%) or supporting recruitment to a research study (32%) **(Figure 13).** Since 2020, the registry has supported 14 surveys. For transparency and to highlight the versatility of the registry, enquiries that the registry has supported are now documented on the registry website.

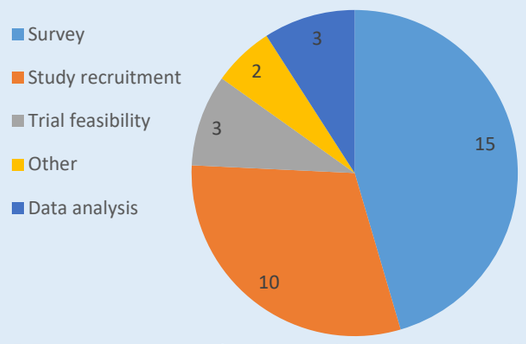

Figure 7. The type and number of registry enquiries supported since 2013

Future Engagement and Collaborations

Ensuring that registry datasets are fit for purpose is vitally important to ensure appropriate and relevant data is available for research. The UK DM Patient Registry is planning a dataset revision process in 2023 in order to align with other TREAT-NMD registries, implement FAIR data principles, and other international standards and agreed data elements. The registry is also keen to support efforts by TREAT-NMD to further update their Core Dataset for DM.

Core Dataset

The UK DM Patient Registry collects all the mandatory and highly encouraged patient-entered items in the TREAT-NMD Core Dataset for DM, based on the agreements made at the 2009 TREAT-NMD/Marigold International Workshop in Naarden. The registry is also a Core Member of TGDOC; this allows the registry to participate in Global Registry Enquiries coordinated by TREAT-NMD.

Global Registry Enquiries

The UK registry has participated in TREAT-NMD Global Registry Enquiries for DM, including providing aggregate data to an industry partner used to successfully support their application for a Rare Paediatric Disease Designation from the FDA .

Other TREAT-NMD collaborations

The UK registry was also pleased to be included in the recent TREAT-NMD poster describing the DM Global Registry Network, presented at the ICMD 2022. Of the 6,472 DM patients reported in the global network, approximately 13% are located within the UK registry.

The TREAT-NMD Core Dataset for Myotonic Dystrophy

The TREAT-NMD Core Dataset for DM includes data elements on age of onset, current best motor function, wheelchair use, myotonia, ventilation support, cardiac disease, dysphagia, and fatigue. The charts below indicate a current snapshot of this data in the UK registry:

Age of onset is included in the registry's clinician-reported questionnaire; this data is currently only available for 35% of patients, however the registry team are investigating strategies to increase clinician data entry levels. Of the reported data, the majority of patients **developed symptoms (24%),** a minority were reported as **congenital (10%)** and a small number are **asymptomatic (1%).** Of the patients who developed DM symptoms, the maximum age of onset reported was 68 years, with the highest frequency of symptom onset reported between the ages of **15 to 29** years.

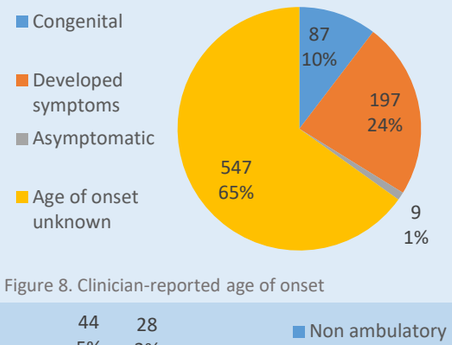

Figure 8. Clinician-reported age of onset

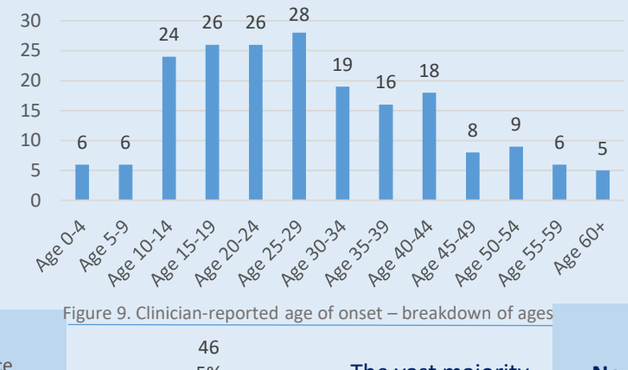

Figure 9. Clinician-reported age of onset – breakdown of ages

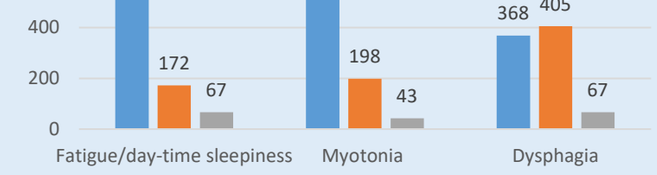

Figure 10. Patient-reported symptoms

Most patients self-report experiencing **fatigue**/day-time sleepiness and **myotonia** (74% and 71% respectively). **Dysphagia** was also reported as a symptom in 44% of patients, however 48% reported no dysphagia symptoms.

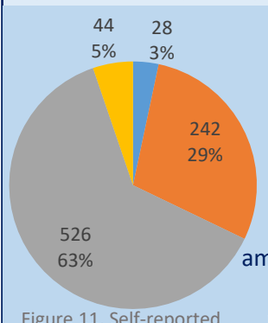

Figure 11. Self-reported current best motor function

Most patients reported their **current best motor function** as either ambulatory (63%) or ambulatory with assistance (29%). A small number of patients reported being non-ambulatory (3%) or did not answer (5%)

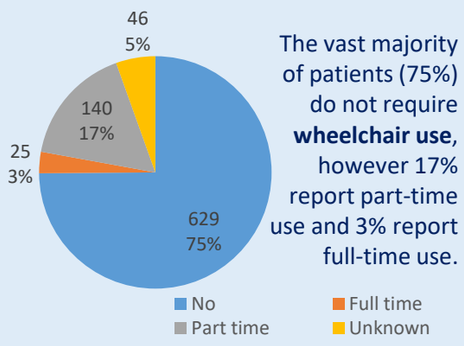

Figure 12. Self-reported wheelchair use

The vast majority of patients (75%) do not require **wheelchair use**, however 17% report part-time use and 3% report full-time use.

Clinician-reported data (available for ~35% of registry patients)

Non-invasive ventilation was reportedly used by 4.2% of patients on a part-time basis. No patients currently report full-time non-invasive ventilation use. Only one patient (0.1%) currently reports using **invasive ventilation,** on a part-time basis.

Pulmonary function testing was reported to have been performed on 22% of patients and had not been performed on 9.4%.

Gastric tube use was reported for only 3 patients (0.4%), whereas no gastric tube use was reported for 32% of patients

Cataract surgery has reportedly been performed on 8.1% of patients; 25% reported no cataract surgery

Acknowledgement to our funders for their continued support and to all the patients and clinicians who continue to participate in the registry.

Contact the UK Registry

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Registry Curator & Project Manager

Dr Chiara Marini Bettolo
Registry Principle Investigator

Registry Website
https://bit.ly/UKDMreg

Email the curator
myotonicdystrophyregistry@newcastle.ac.uk

Use the registry data in your research
https://bit.ly/dmenquiry

The UK DM Patient Registry continues to be a versatile, cost-effective research tool that has helped facilitate a range of studies and advance DM research around the world. Additional work is planned to update the registry questionnaires, improve engagement with more doctors in the UK and increase the reporting of genetic information on the registry. As well as supporting research projects, the registry continues to develop new and engaging communication materials for the DM community and plans to further capture the patient voice in the development of new materials.