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

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Thanks to our registry funders:

Muscular Dystrophy UK – Fighting muscle-wasting conditions

Cure DM – Support ♥ Awareness ♥ Research

Myotonic Dystrophy Support Group

The John Walton Muscular Dystrophy Research Centre

Background

The UK Myotonic Dystrophy (DM) Patient Registry is a patient self-enrolling online database collecting clinical and genetic information about all types of DM. The registry was established in May 2012, is supported by Muscular Dystrophy UK, CureDM and the Myotonic Dystrophy Support Group, and coordinated by Newcastle University.

Aims

The registry aims to better characterise and understand the natural history of DM, and implementation of standards of care. It also facilitates academic and clinical research, and disseminates information on studies and research advancements to participants.

Method

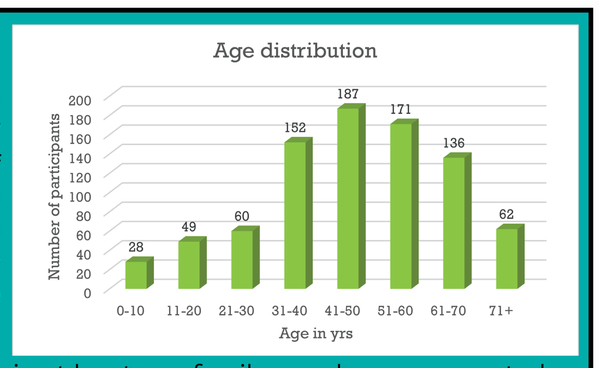
The registry captures longitudinal, self-reported patient questionnaire data via a secure online portal. Neuromuscular specialists involved in patient care can also be invited to provide additional clinical or genetic information as required. The registry is a Core Member of the TREAT-NMD Global Registries Network for Myotonic Dystrophy, collecting their standardised core dataset and contributing to global data enquiries.

Results

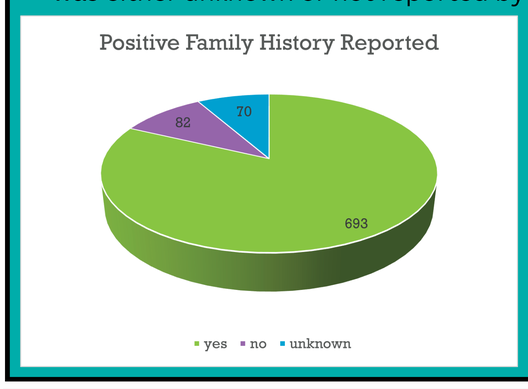
As of April 2024, there were 845 active patient registrations. Data is also available for an additional 171 patients who are deceased, unresponsive or not based in the UK (their data is not included here). For those reporting a clinical diagnosis, 76.9% have DM1, 4.4% have DM2, and 10% have congenital DM. Genetic confirmation has been reported by 42.1% of participants. In addition to collecting specific genetic data inputted by clinicians, the registry is now able to receive digital copies of patient's genetic reports directly via a secure upload portal. The registry has supported many enquiries to date, recent examples include providing anonymous patient-level data with genetic test results and symptom onset age to support an academic research project, aggregated data for a global study into DM2 prevalence, and several research surveys.

Demographics

The ages of registry participants range from 0 to 86 years, with an average age of 46.7 years. Adults (age 18-64) comprise 76.8% of the participants, with elderly (age 65+) making up 15.7%, and paediatric (under 18) totalling 7.5% of participants. Sex is fairly evenly distributed; 48.2% of patients are male and 51.8% are female.



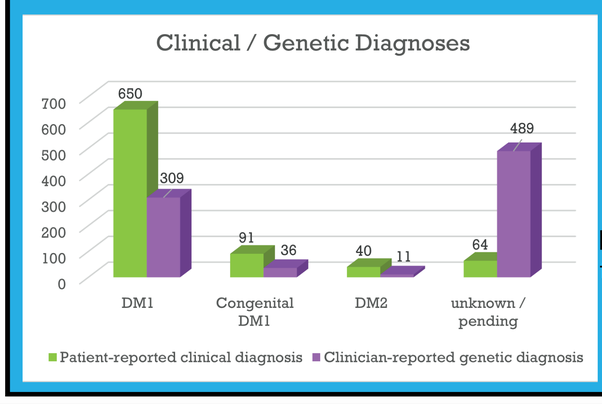
A positive history of Myotonic Dystrophy in at least one family member was reported by 82% of patients, with 9.7% reporting no known family history. Family history of DM was either unknown or not reported by 8.3% of participants.

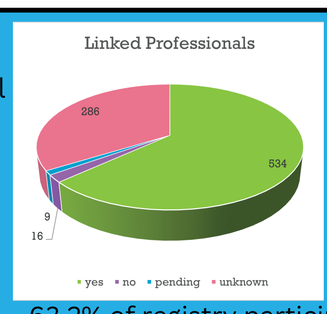


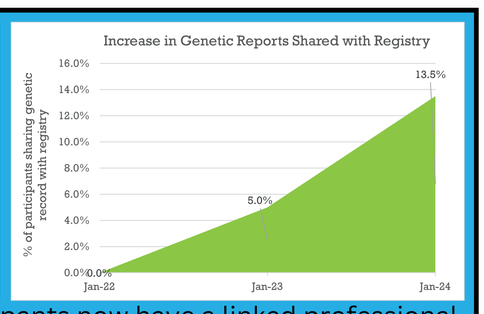
The majority of registry participants have reported their ethnicity as Caucasian (90.9%). Other ethnicities reported were Mixed (2.0%), 'Other' (0.8%), Asian (0.6%) and Black African (0.4%). 5.3% have not yet reported their ethnicity. More granularity in ethnicity reporting options will be included in the next registry dataset update in order to better capture this data for analysis, and also to improve diversity within the registry.

Diagnoses

92.4% of participants reported their clinical diagnosis, with 7.6% awaiting confirmation. This option is often chosen by participants who are unsure whether they have DM1 or DM2, and is updated once genetic confirmation of diagnosis is shared with the registry. Genetic confirmation of diagnoses is now available for 47.5% of the self-reported DM1 patients, 39.6% for congenital, and 27.5% for DM2.







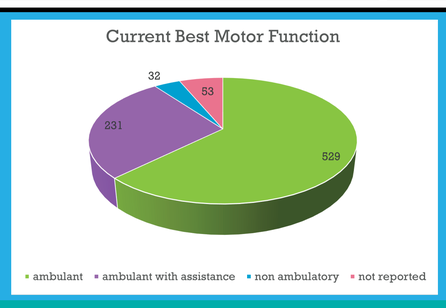
63.2% of registry participants now have a linked professional user (neuromuscular consultant, genetic counsellor, physio etc.) to verify patient-entered data, provide clinical data from medical records, and confirm genetic data reporting. 1.9% report they do not currently see a specialist, 1.1% have a professional user with a pending invitation, and 33.8% have not yet provided details of their specialist.

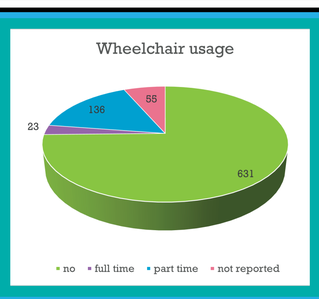
The registry implemented a new feature in 2022 for participants to upload their genetic test results for the curator to enter as clinical data, increasing the amount of genetic data available for research purposes in the registry. This is proving to be successful, with 13.5% of all registry participants having shared their full genetic report to date.

Patient-reported clinical features

The registry collects patient-reported data for many of the common clinical features of Myotonic Dystrophy.

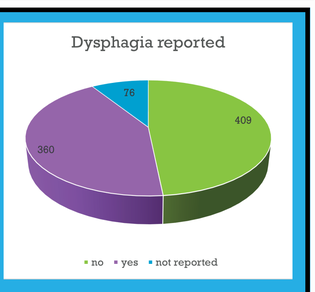
Most patients reported their current best **motor function** as either ambulatory (62.6%) or ambulatory-assisted (27.3%). A small number of patients reported being non-ambulatory (3.8%), and motor function was not reported by 6.3%

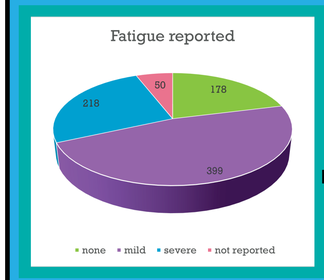




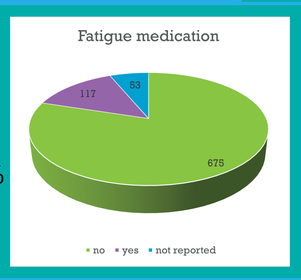
The majority of registry participants do not currently require the use of a **wheelchair** (74.7%), however 16.1% report part-time use and 2.7% report full-time use. This data is not yet available for 6.5% of participants.

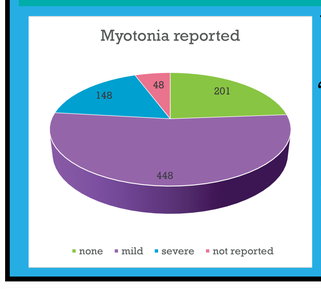
Almost half of all registry participants report no **dysphagia** (48.8%), however 42.6% do report dysphagia symptoms. This data is not reported for 9% of participants.



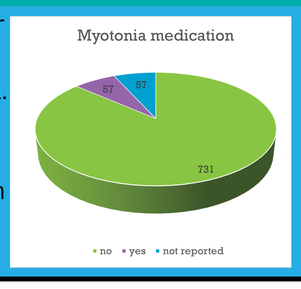


Almost three-quarters of patients (73%) reported experiencing **fatigue**. This was reported as 'mild' by almost half of the registry participants (47.2%), and 'severe' by around a quarter (25.8%). Most (79.9%) do not use any medication to manage fatigue - only 13.8% report use of fatigue medication. A fifth of all registry participants (21.1%) reported experiencing no fatigue.



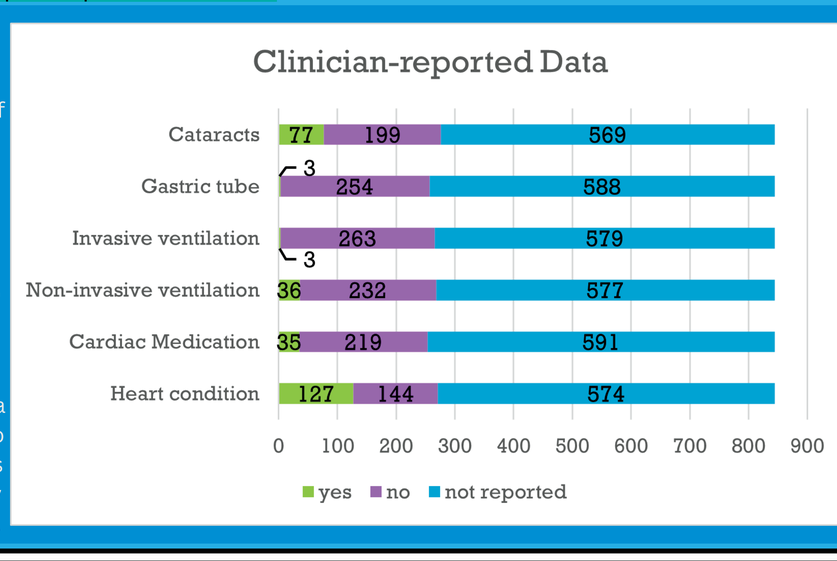


The majority of patients (70.5%) reported **myotonia**. Over half of the registry participants (53%) describe this as 'mild', and almost a fifth (17.5%) report 'severe' myotonia. Almost a quarter of participants (23.8%) report no myotonia symptoms. Use of medication to manage their myotonia was only reported by 6.7% of participants, with the vast majority (86.5%) reporting taking no medication for this reason. 6.7% did not answer.



Clinician-reported data

Clinical data can be reported to supplement patient-entered data if the patient nominates their neuromuscular specialist. The figures on clinical data reported here should be considered as low estimates, as not all patients provide details of their specialists, and some nominated clinicians do not report clinical data regularly. The registry team are working on overcoming barriers to clinical data entry and investigating how best to support professional registry users in order to increase the availability and completeness of clinical data .



Conclusion

The UK Myotonic Dystrophy Patient Registry is one of the largest national DM patient registries globally and is an example of a versatile, cost-effective research tool, helping to facilitate and advance a wide range of DM research. The genetic report upload feature is shown to be significantly improving the genetic information available on the registry, alongside the increase in neuromuscular specialists signing up as professional users. The patient and clinical questionnaires will be reviewed and updated in the near future, and data linkage is planned between the registry and the Newcastle Research Biobank for Rare and Neuromuscular Diseases which will enable more data to be available to facilitate research into DM. Additional work around patient engagement and promotion of the registry to neuromuscular specialists is ongoing to increase the number of patients aware of and signing up to the registry, and efforts are required to increase the diversity of the registry population and improve the registry system to provide a better user experience.

Meet the Registry Team

Ms Helen Walker
Registry Curator & Project Manager

Visit the registry website

Email the registry curator

Learn about the other JWMDRC Registries

Registry Website: bit.ly/UKDMreg

Dr Chiara Marini Bettolo
Registry Principle Investigator