

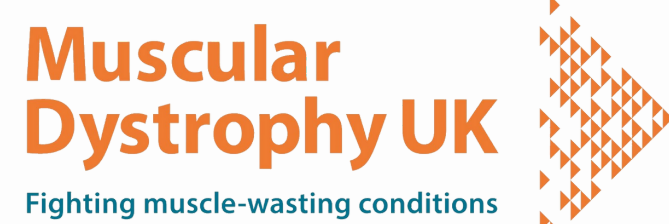
# The UK Myotonic Dystrophy Patient Registry: Empowering Clinical Research and Patient Voice with an Effective Translational Research Tool

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10. Sheffield Teaching Hospitals NHS Foundation Trust, Sheffield, UK
11. Myotonic Dystrophy Support Group, Nottingham, UK

Thanks to our registry funders:



## 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, Cure-DM and the Myotonic Dystrophy Support Group, and coordinated by Newcastle University.

## Aims

The registry facilitates academic and clinical research, enables better characterisation and understanding of DM, and disseminates information relating to upcoming studies and research advancements to participants.

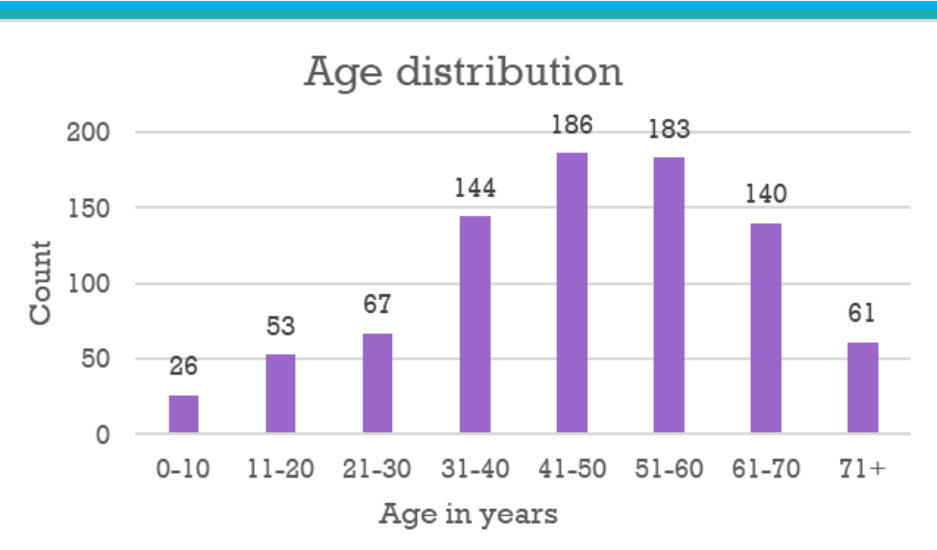
## Method

The registry captures longitudinal, self-reported data via a secure online portal. Where specialised clinical or genetic information is required, neuromuscular specialists involved in patient care are invited to provide additional 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.

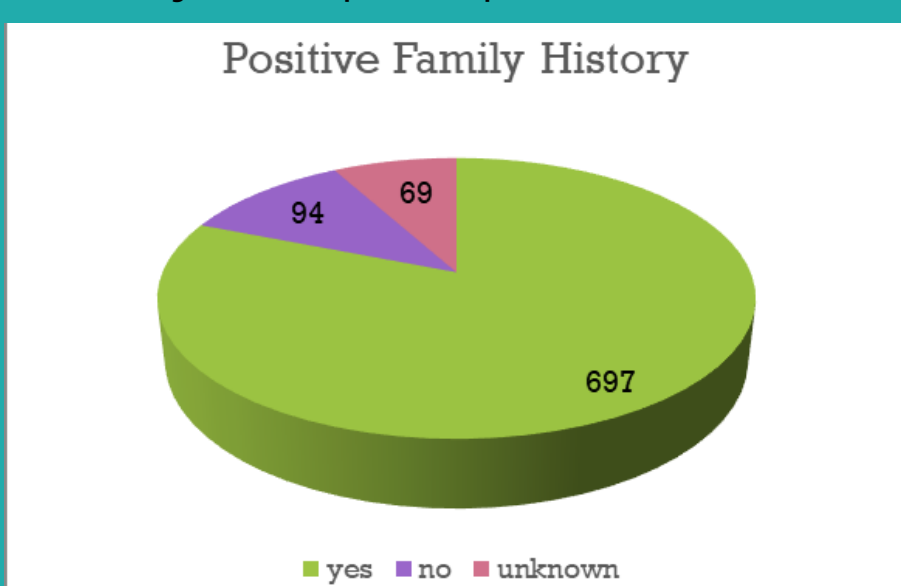
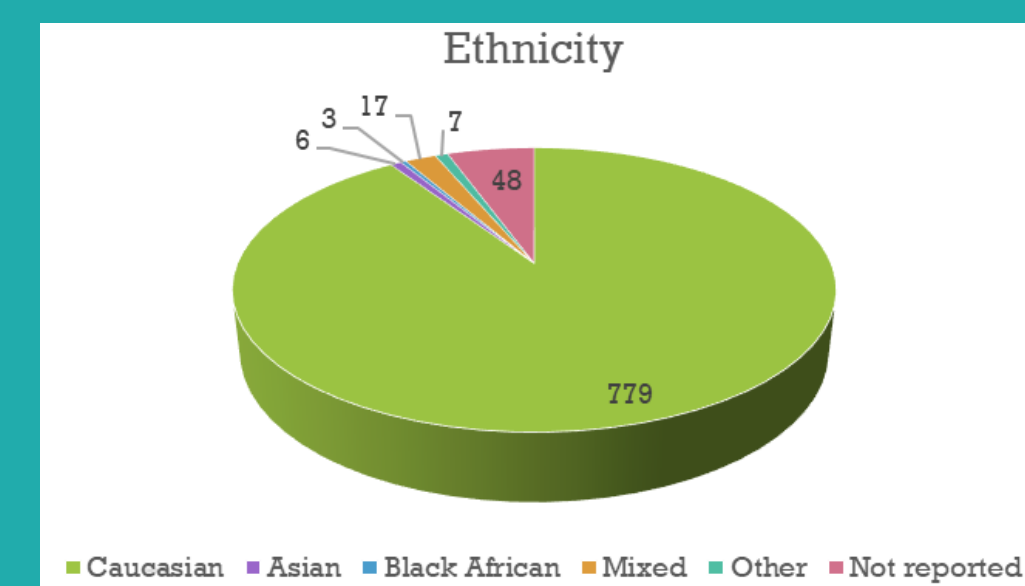
**Results:** As of August 2023, there were 860 active, UK based patient registrations. Data is also available for an additional 141 patients who are deceased, unresponsive or not based in the UK (their data is not included here). For those reporting a clinical diagnosis, 75.1% have DM1, 4.3% have DM2, and 9.8% have congenital DM. Genetic confirmation has been reported by 39.1% of patients. 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 32 registry enquiries to date, recent examples including a global study into DM2 prevalence, a survey on UK service provision, and various surveys capturing information on patient preferences, dysphagia, pregnancy, sleep, and the patient/caregiver experience.

## Demographics

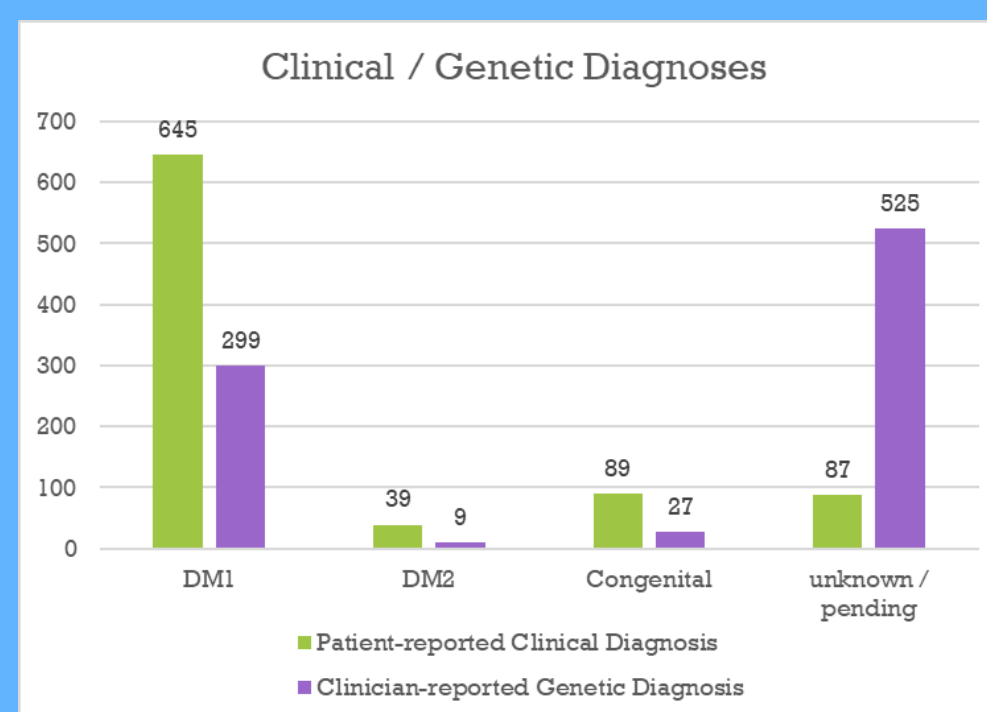
The ages of registry participants range from 0 to 85 years, with an average age of 46.8 years. Adults (age 18-64) comprise 76.6% of the participants, with elderly (age 65+) making up 165%, and paediatric (under 18) totalling 7.3% of participants. Sex is evenly distributed; 49% of patients are male and 51% are female.



The majority of registry participants reported their ethnicity as Caucasian (90.6%). Other ethnicities reported were Mixed (2.0%), 'Other' (0.8%), Asian (0.7%) and Black African (0.3%). 5.6% did not report their ethnicity. A history of DM in at least one family member was reported by 81% of patients, with 10.9% reporting no known family history. Family history of DM was either unknown or not reported by 8% of participants.

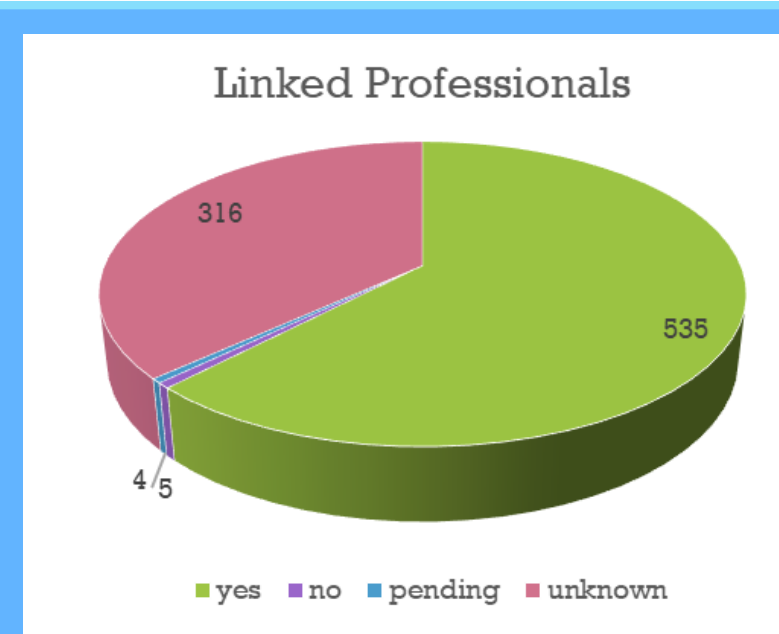


The most common patient-reported clinical diagnosis is DM1, affecting 75.1% of participants. Congenital DM is reported by 9.8%, 4.3% report DM2, and 9.5% are awaiting confirmation, or have not yet reported a diagnosis. This option is often chosen by participants who are unsure whether they have DM1 or DM2, and is updated once genetic confirmation of diagnosis is shared with the registry.



## Diagnoses

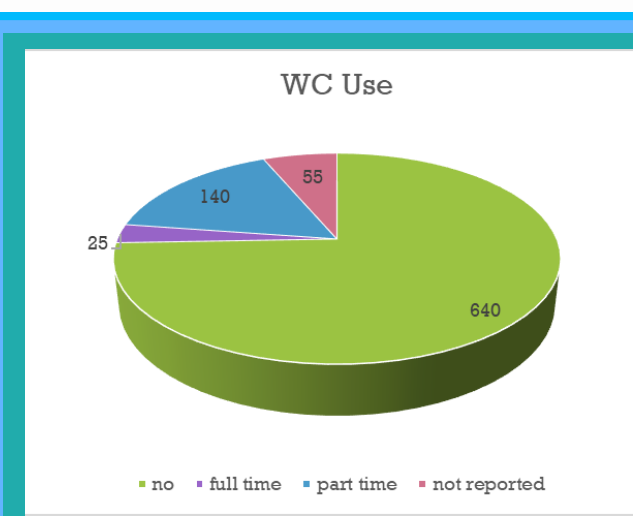
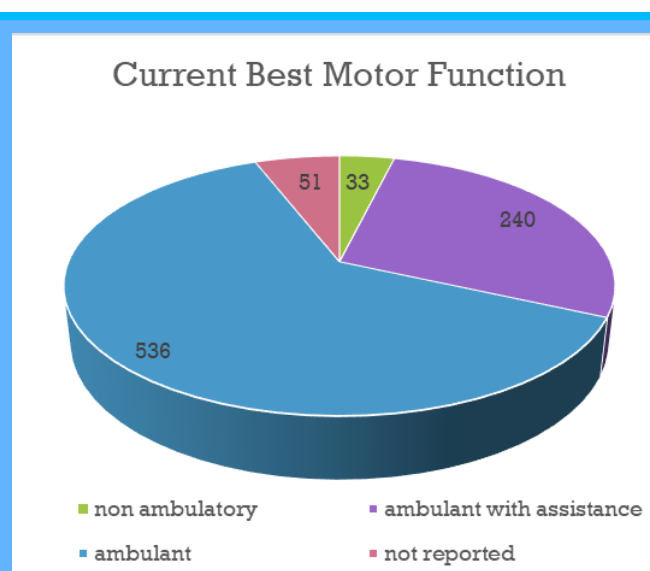
Genetic report upload is a new feature of the registry!



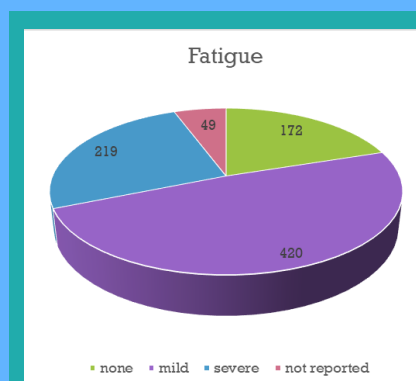
62% of registry participants now have a linked professional user (neuromuscular consultant, genetic counsellor, physio etc.) to verify patient-entered data, provide clinical data from medical records, and confirm genetic data reporting. 0.6% report they do not currently see a specialist, 0.5% have a professional user with a pending invitation, and 37% have not answered this question. The registry implemented a new feature in 2022 whereby participants can upload a copy of 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.

## Clinical features

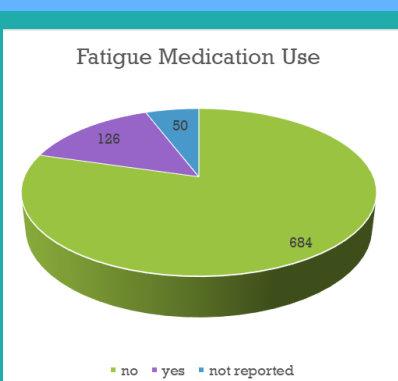
Most patients reported their current best motor function as either ambulatory (62.3%) or ambulatory-assisted (27.9%). A small number of patients reported being non-ambulatory (3.8%), and motor function was not reported by 5.9%



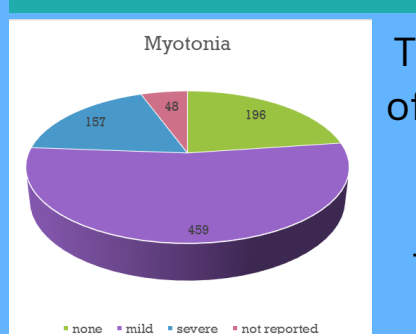
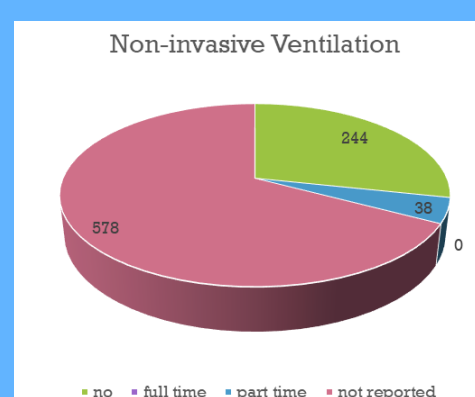
The majority of registry participants do not currently require the use of a wheelchair (74.4%), however 16.3% report part-time use and 2.9% report full-time use. This data is not yet available for 6.4% of participants.



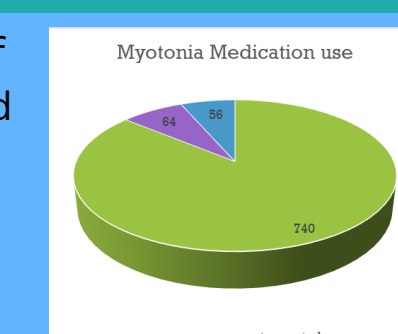
Almost three-quarters of patients (74.3%) reported experiencing fatigue. This was reported as 'mild' by almost half of the registry participants (48.8%), and 'severe' by around a quarter (25.5%). Most (79.5%) do not use any medication to manage fatigue - only 14.7% report use of fatigue medication. A fifth of all registry participants (20%) reported experiencing no fatigue.



Non-invasive ventilation was reported for only 4.4% of patients, all of whom are reported to use this part-time. Similarly, only 0.3% are reported to use invasive ventilation on a part-time basis. These figures should be considered as low estimates, as medical professionals have not yet reported this data for 67% of patients.



The majority of patients (71.6%) reported myotonia. Over half of the registry participants (53.4%) describe this as 'mild', and almost a fifth (18.3%) report 'severe' myotonia. Use of medication to manage their myotonia was only reported by 7.4% of participants, with the vast majority (86%) reporting taking no medication for this reason. 6.5% did not answer.



The registry also collects data on the following:

- Dysphagia
- Cardiac involvement
- Medication use
- Surgery and other interventions
- Use of gastric tube
- Cataracts
- Pulmonary function testing
- ECG results

Clinical data is reported by patient's nominated specialist,

or a member of their team. The registry team are working on overcoming barriers to data entry and investigating how best to support professional registry users in order to increase the availability and completeness of clinical data available in the UK DM Patient Registry.

## Meet the Registry Team



**Ms Helen Walker**  
Registry Curator & Project Manager



**Dr Chiara Marini Bettolo**  
Registry Principle Investigator



Registry Website  
[bit.ly/UKDMreg](https://bit.ly/UKDMreg)

Use the registry data in your research  
[bit.ly/dmenquiry](https://bit.ly/dmenquiry)



Email the registry curator



Learn about the other JWMDRC Registries

## Conclusion

The UK 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 new genetic report upload feature is shown to be improving the genetic information available on the registry, alongside the increase in neuromuscular specialists signing up as professional users. There are plans to review and update the patient and clinical questionnaires in the near future, and data linkage plans 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 are 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.