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

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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 the John Walton Muscular Dystrophy Research Centre (JWMDRC) at Newcastle University.

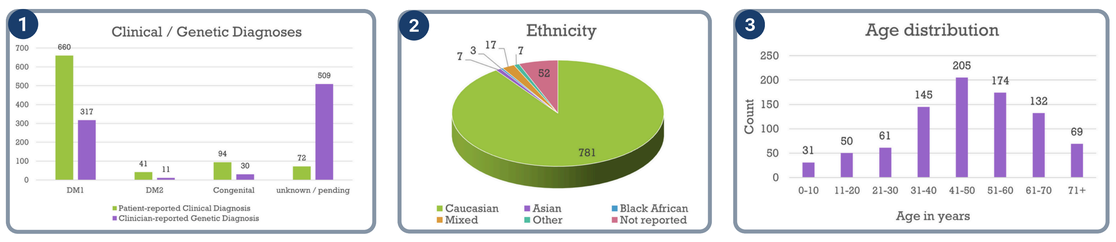
Aims

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 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.

Figures



Results

As of September 2024, there are 867 active patient registrations in the registry. Data from an additional 141 deceased, unresponsive or inactive patients is not included in this analysis. For those reporting a clinical diagnosis, 76.1% have DM1, 4.5% have DM2, and 10.4% have the congenital form [*fig.1*]. Genetic confirmation of the condition has been received for 40% of active patients to date. Over 90% of participants report their ethnicity as Caucasian, 2% as mixed, and 6% did not answer [*fig.2*]. The average age of participants is 47 years, with a range between 1 to 68 years [*fig.3*], and sex is fairly evenly represented (52% female, 48% male). Fatigue and myotonia are the most common symptoms, reported by 72.4% [*fig.4*] and 70% [*fig.5*] of patients respectively. Dysphagia was reported by 42.4% *[fig.6*]. Only 8.7% of patients with myotonia and 18.2% with dysphagia reported medication use for symptom management. Most patients reported their current best motor function as ambulant (62%) or ambulant with assistance (27%) [*fig.7*]; 16% reported using a wheelchair part-time and 2.5% full-time [*fig.8*]. Part-time non-invasive ventilation was reported for only 4.4% of patients [*fig.9*] and no full-time or invasive ventilation is currently reported, however ventilation status is currently only entered by clinician users and only available for 31% of patients.

Conclusions

One of the largest national DM patient registries and an example of a versatile, cost-effective research tool helping to facilitate and advance a wide range of research, the registry has supported 38 registry enquiries. Recent examples include a global study into DM2 prevalence, recruitment support for two dysphagia studies, and surveys on service provision, patient preferences, and patient/caregiver experiences. Additional work continues to be done to improve reporting of genetic information on the registry, and a dataset expansion project is currently underway to ensure data collected is relevant and useful. New data to be collected will include Patient-Reported Outcome Measures (PROMs), trial readiness, and access to care questions, and Privacy Protecting Record Linkage (PPRL) items to support anonymous data sharing. Ethnicity and other demographic questions will be refined in the updated dataset to allow for more accurate and detailed cohort information, and to inform PPIE activities to increase diversity, increase awareness of the registry in under-represented groups, and remove any barriers to participation. The registry is also transitioning to a new bespoke software platform incorporating automatic data validation and logic, data dashboards, analysis tools, and an improved user experience for patients and clinicians. These developments will support further patient recruitment and ensure registry data available to support research into DM is of the highest quality.

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Meet The Team

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Registry Manager

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