The UK Myotonic Dystrophy Patient Registry: An Important Tool Linking Patients to National and International Research Projects

Background

- The UK Myotonic Dystrophy Patient Registry (https://www.dm-registry.org.uk/) is a patient self-enrolling online database, collecting clinical and genetic information about both myotonic dystrophy type 1 (DM1) and myotonic dystrophy type 2 (DM2).
- Established in May 2012.
- Supported by Muscular Dystrophy UK (MDUK) and the Myotonic Dystrophy Support Group (MDSG), assisted by the TREAT-NMD Alliance (www.treat-nmd.org) 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 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 clinicians.
- Patient reported outcomes are entered into a secure portal, combined with clinician verified clinical and genetic details.
- The dataset collected within the registry includes all mandatory and highly encouraged items agreed at the 2009 TREAT-NMD and Marigold Foundation workshop held in Naarden.
- This includes patient reported data items such as, clinical diagnosis, wheelchair use, myotonia, fatigue/daytime sleepiness, current best motor function, and doctor reported items such as heart condition, electrocardiogram, echocardiogram medication, ventilation, age of onset and genetic confirmation.
- The registry can support researchers and industry on various projects by facilitating study/survey recruitment or by providing de-identified patient data.

Results

- As of April 2021, there were 808 participants enrolled on the UK Myotonic Dystrophy Patient Registry. This includes 409 female and 391 male participants, with an average age of 44.9 years old (range of 0 – 83 years).

Fatigue/day-time sleepiness and myotonia

- Eleven percent of patients report medication use to treat myotonia (Figure 7), with myotonia as the most commonly used medication (34%).
- Twenty percent of patients report medication use for fatigue/day-time sleepiness (Figure 8), with modafinil as the most commonly used medication (87%).

Doctors have reported on the cardiac section of the registry’s doctors form in 35% of patients. Almost half (47%) of these patients have a heart condition (Figure 9), with a mean age of onset as 36.3 ± 18.3 years.

The heart conditions reported by doctors were:
- Conduction block in 44% of cases
- Arrhythmia or conduction block in 33% of cases
- Arrhythmia in 10% of cases
- Other in 8% of cases

Medication use for a heart condition was reported in 22% of cases.

An electrocardiogram was reported for 243 patients (30%) on the registry. The mean PR interval was 187.3 ± 38.2 milliseconds (ms), and the mean QRS duration was 107 ± 31.6 ms.

An echocardiogram was provided for 129 patients (16%) and the mean left ventricular ejection fraction (LVEF) was 61.3 ± 10.5%. The below LVEF ranges were recorded in 100 patients:
- Hyperdynamic (LVEF >70%) in 10 cases (10%)
- Normal (LVEF 50%-70%) in 84 cases (84%)
- Mild dysfunction (LVEF 40%-49%) in 4 cases (4%)
- Moderate dysfunction (LVEF 30%-39%) in 2 cases (2%)

The results of pulmonary function testing were reported in 22% of patients, with forced vital capacity (FVC) recorded as (Figure 10):
- Normal in 53% of cases
- Moderate in 13% of cases
- Moderately severe in 13% of cases
- Severe in 25% of cases

From the available data of 17 studies supported by the registry, recruitment has ranged from 17%-84%, with a mean recruitment of 55% when supporting clinical trials and research studies.

Registry utilisation in research

To date the registry has supported 31 enquiries from industry, academics, clinicians and patient organisations. Most registry enquiries involve having online survey distribution (48%) or supporting clinical trial and research student recruitment (32%). Since 2020, the registry has supported 11 surveys, and 1 confidential industry enquiry.

For transparency and to highlight the versatility of the registry, enquiries that the registry has supported are now documented on the registry website.

Conclusion

- 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 continues to be done to improve engagement with more doctors in the UK and the reporting of genetic information on the registry. There are also future data linkage plans between the registry and the Newcastle Research Biobank for Rare and Neuromuscular Diseases. As well as supporting research projects, the registry continues to develop new and engaging communication materials for the UK DM community, and plans to further capture the patient voice in the development of new materials.

The registry has also been involved in 28 publications most of which include studies where the registry has supported recruitment.

These are also now documented on the registry website and are routinely updated.