Expanding the UK Myotonic Dystrophy Patient Registry Dataset Improving Data Collection and Amplifying 'Patient Voice'

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Background

The UK Myotonic Dystrophy Patient Registry

Since 2012, the UK **Myotonic Dystrophy** (DM) Patient Registry has collected the TREAT-NMD Core Dataset for DM, a minimal list of data items agreed at a 2009 ENMC workshop. The dual-reported registry c**ollects both patient-entered and clinical data** using this dataset, creating a valuable repository of **longitudinal data** on their national patient cohort. The registry is coordinated by the John Walton Muscular Dystrophy Research Centre (JWMDRC) at Newcastle University and contributes deidentified data with TREAT-NMD Global Registries Network for **global research enquiries**, and provides **data reports** and **recruitment support** to academics, healthcare professionals and industry. A need was identified to **update the registry dataset** to ensure data collection remains **relevant to current research requirements,** and the registry is able to **facilitate regulatory approval projects**.

TREAT-NMD Core Dataset for DM

The 2009 standardised core dataset contains mandatory patient-entered questions on diagnosis, family history and ethnicity, and mandatory longitudinal questions on motor function, wheelchair use, symptoms and medication. Optional questions on pregnancy history and children are also included. The clinician-entered section can be completed by patients' healthcare professionals and includes cardiac information, ECG and Echo results, pulmonary function testing and ventilation support, feeding tube use, cataract surgery, age of symptom onset, and some genetic test results (if available).

Aims

In 2023, the UK DM Registry identified the need to **expand and refine the questionnaires** within the registry dataset, and a Working Group (WG) was established comprising academic, genetic, physiotherapy, clinician and patient advocate experts from the registry's Steering Committee with the following aims:

- Review dataset to ensure data collection remains **relevant** and **appropriate** to the UK's patient and research communities.
- Balance research **data requirements** with **patient burden** of data entry
- Utilise **new platform functionality** for more complex Q&A types, (branching logic, conditionals, data validation, scoring etc.)

Methodology

The Registry Dataset WG assessed data items and questionnaires in the following ways:

Review	Refine	Expand
Relevance	*Definition*	*Add PROMs*
Currentness	*Regionalise*	*Data validation rules*
Completeness	*Question type*	*Device usage episodes*
Mandatory items	*Conditional display*	*Add response specificity*
Align with other datasets	*Answering options*	*Adult / paediatric sections*

FAIR data standards are also incorporated into the new dataset, making it widely accessible and interoperable. Patient-friendly supplementary language was added wherever advised by WG patient representative, and questionnaires are **personalised** based on user and patient profile in the new registry platform, ensuring only relevant questions are presented to adult patients, and parents/guardians of paediatric patients.

Conclusion

The UK DM Patient Registry's expanded dataset is **a broad, flexible resource to capture real-world data.** The dataset has been **aligned** with other relevant data collection activities, and mandatory questions have been carefully minimised to balance data requirements with the patient burden of data entry. The registry's transition to bespoke registry platform allows **direct management of registry questionnaires** and ensuring new data collection projects can be setup **quickly and efficiently.**

Results

The expanded dataset was **incorporated into the new data collection platform** that the registry adopted in April 2025. Details will be made available on the registry website and the dataset can be adopted by other national DM registries and data collection initiatives to **encourage harmonisation and alignment** of data collection.

Overview of New Patient and Medical Questionnaires

Patients have the option to **nominate a named neuromuscular specialist** to complete their Medical Questionnaire and can also complete this themselves and **upload medical documentation** for data verification. Clinical users can also contribute information to **a Genetic Test Results** Form, or patients can upload their genetic report themselves. The Patient Questionnaire includes newly added sections on **trial participation preferences**, **PROMs**, and opportunities to share **experiences of specialist care**. The following sections appear in the new registry questionnaires:

Patient Questionnaire
for patient/caregiver data entry only

Diagnosis & Demographics
- Clinical diagnosis
- Genetic report status
- Affected family
- Ethnicity

Access to Care
- NMD specialists last seen
- NMD care site
- Dates, frequency & appointment type
- Access to counselling
- Care funding & management
- Social care child & adult
- Care comments

Access to Research
- Current & previous trial participation
- Trial preferences
- Type, interventions, rating

Patient Reported Outcome Measures (PROMs)
- SOFT Scale
- Myotonia Behaviour Scale Score
- Epworth Sleep Scale Score
- Fatigue Severity Scale Score
- Visual Analogue Fatigue Scale (VAFS)
- Patient Global Impression (PGI)
- Patient Global Impression of Change (PGI-C)
- DM1-ACTIVc Score

Mobility
- WC Use
- Status, dates, type
- Additional Aids

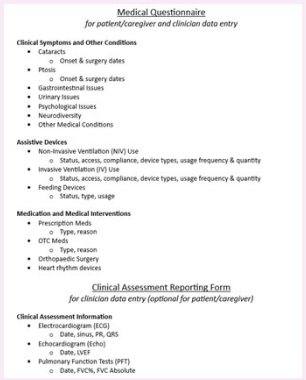
Symptoms
- Symptoms & Onset
- Dysphagia & Onset
- Grip Strength

Fertility and Family Planning
- Family planning
- Fertility Issues
- Pregnancies & outcomes

Physiotherapy
- Status, access, dates
- Main symptoms and benefits

Speech & Language Therapy
- Status, access, dates
- Main symptoms and benefits

Occupational Therapy
- Status, access, dates
- Main symptoms and benefits



Medical Questionnaire
for patient/caregiver and clinician data entry

Clinical Symptoms and Other Conditions
- Cataracts
  - Onset & surgery dates
- Ptosis
  - Onset & surgery dates
- Gastrointestinal Issues
- Urinary Issues
- Psychological Issues
- Neurodiversity
- Other Medical Conditions

Assistive Devices
- Non-Invasive Ventilation (NIV) Use
  - Status, access, compliance, device types, usage frequency & quantity
- Invasive Ventilation (IV) Use
  - Status, access, compliance, device types, usage frequency & quantity
- Feeding Devices
  - Status, type, usage

Medication and Medical Interventions
- Prescription Meds
  - Type, reason
- OTC Meds
  - Type, reason
- Orthopaedic Surgery
- Heart rhythm devices

Clinical Assessment Reporting Form
for clinician data entry (optional for patient/caregiver)

Clinical Assessment Information
- Electrocardiogram (ECG)
  - Date, sinus, PR, QRS
- Echocardiogram (Echo)
  - Date, LVEF
- Pulmonary Function Tests (PFT)
  - Date, FVC%, FVC Absolute

Data Item Comparison Examples

The following examples demonstrate how the five mandatory data items in the 2009 TREAT-NMD Core Dataset for DM are collected in the new dataset:

Old Dataset		New Dataset	
What is your diagnosis, according to your doctor?	Congenital Myotonic Dystrophy DM1 DM1 asymptomatic mutation carrier Other Unknown	What is your diagnosis, according to your doctor?	Myotonic Dystrophy (Unsure of subtype) DM1 - Congenital onset (symptoms before 4 weeks) DM1 - Childhood onset (symptoms before age 18) DM1 - Adult onset (symptoms age 18+) Myotonic Dystrophy Type 2 Other I don't know
What is your genetic test result?	DM1 mutation (triplet repeat expansion) – provide date, lab, method & repeat size Other mutation:..... Result pending I have not been tested	Combined with **Clinical Diagnosis** & **Genetic Test Report Form** (Clinical / Registry use only)	
Which of the following options describes the best motor function you are currently able to achieve?	Ambulatory (unassisted) Ambulatory (assisted) Non-ambulatory	Which of the following options describes your current best motor function? *[in SOFT]*	Unable to sit upright in wheelchair (reclined or no longer able to use WC) Unable to walk, mainly electric wheelchair user Unable to walk, mainly manual wheelchair user Walks with aids (stick, frame, person) Walking speed, no aids, no acceleration Picks up speed, accelerates Able to run
Do you use a wheelchair?	No Part-time (age...) Full-time (age ...)	Have you ever used a wheelchair due to your Myotonic Dystrophy?	Yes currently / Yes previously / No never Start date / frequency of use Type of WC / how often / circumstances / start date / continuous use / episode reporting

Data Sharing & Future-proofing

As the number of **clinical trials** arriving in the UK increases, the ability to collect data on **potential treatments or disease modifying therapies** becomes crucial. The expanded dataset has been developed to allow the registry system to run studies to provide real-world data to support **post-marketing surveillance** activities. It also enables the creation of **Privacy Preserving Record Linkage** keys to enable patient-level data to be anonymously shared and linked with other data sources, **greatly increasing the useability and value** of the data for research.

We gratefully acknowledge our registry supporters

We are grateful to all registry participants, steering committee members, patient representatives, wider study team, and all supporters of the UK DM Patient Registry.

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