New developments and data highlights in the international myotubular and centronuclear myopathy patient registry

An international research database to accelerate the pace of research and treatment.

Bullivant, J., Lennox, A., Hollander, A., Saegert, C, Lynch, O, Moat, D, Graham, R, Schara-Schmidt, U, Bönnemann, C, Jungbluth, H, Buj-Bello, A, Dowling, J, Marini-Bettolo, C

The Myotubular and Centronuclear Myopathy Patient Registry

Find out more: **www.mtmcnmregistry.org**
Contact us: **mtmcnmregistry@newcastle.ac.uk**

WHAT IS THE REGISTRY?

The MTM and CNM Patient Registry is an open-ended research database collecting clinical and genetic data. Participants log in to complete the questionnaire, upload genetic or biopsy reports, and nominate their doctor to provide some clinical data on their behalf. They are asked to check and update their information every 6 months.

The purpose of the registry is to:

- Identify participants for clinical research.
- Understand the epidemiology, genetics, symptoms and severity of the conditions.
- Support existing and future research and inform better standards of care.
- Provide a communications interface between patient and research communities.

Inclusion criteria:

- ✓ Living or deceased individuals diagnosed with MTM or CNM (confirmed via genetic testing or biopsy).
- ✓ Female carriers of X-linked MTM.

International coverage:
Questionnaires are available in English, German, French, Spanish, Italian, Polish, Hindi, Dutch and Brazilian Portuguese, with one more language to follow in 2022.

COHORT DESCRIPTION

On 17-06-2022 the registry had **429** participants (**376** living and **53** deceased). The **376** living participants comprised **220** male and **157** female (of which **86** registered as XLMTM carriers and **71** registered as patients). The **53** deceased participants comprised **51** male and **2** female.

Of the **323** registrations (**282** living and **41** deceased) reporting a causative gene, **193 (60%)** had been genetically confirmed by the registry and this work is ongoing.
53 countries were represented, with the largest cohorts being **UK & Ireland** (107), **United States** (102) and **Germany** (37).

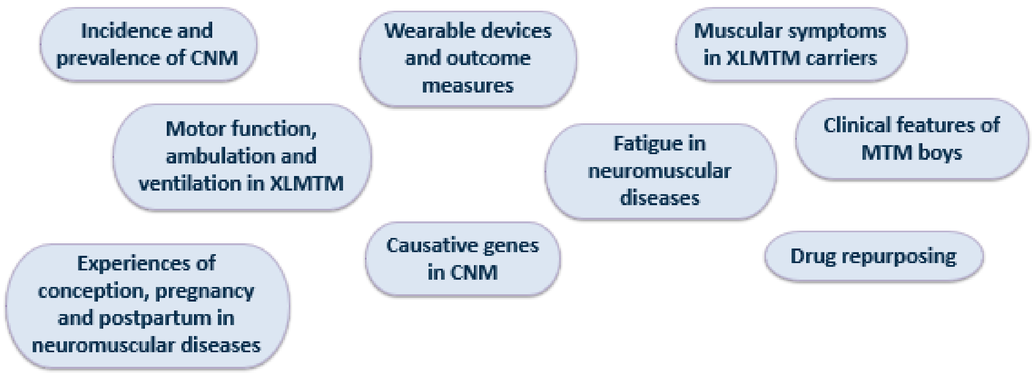
DATASET	
Demographics and personal data	Genetic and/or biopsy report
Clinical diagnosis	Neuromuscular examination dates
Doctor's details	Motor functions
Wheelchair use	Respiratory function and ventilation use
Scoliosis surgery	Chest infection medication
Heart function (ECHO/ECG results)	Feeding function
Other illnesses (carriers only)	Family history
Clinical trial participation	Other registries or studies joined

HOW IS THE REGISTRY USED?

Data are de-identified and made available to researchers or companies (if approved by the Registry Steering Committee).

	2019	2020	2021	2022
Number of registry enquiries	2	2	4	2

Research supported so far includes (but is not limited to):



CLINICAL FEATURES

Chart 1. Best reported motor function

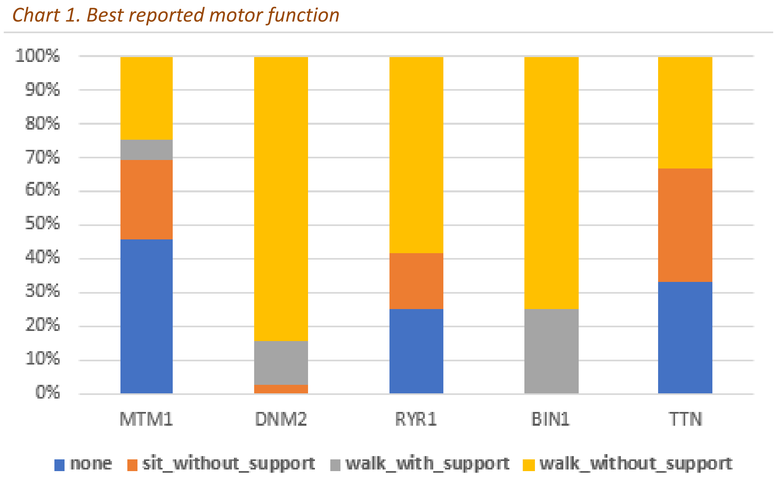


Chart 2. Ventilation required at birth

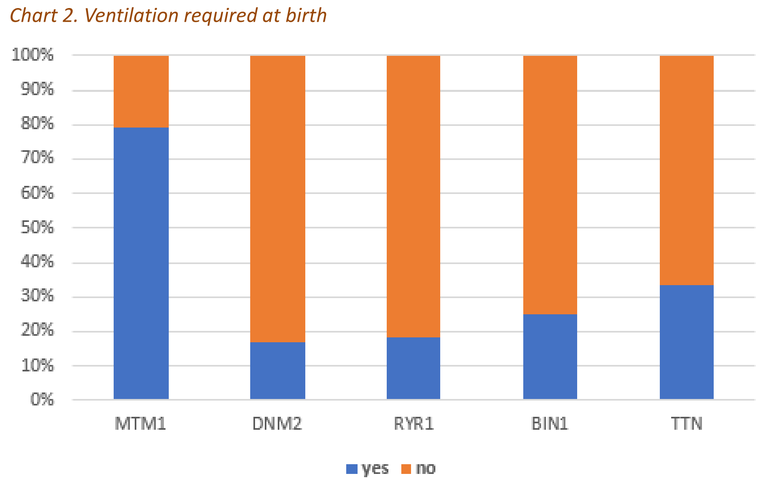


Chart 3. Abnormal electrocardiogram (ECG) results

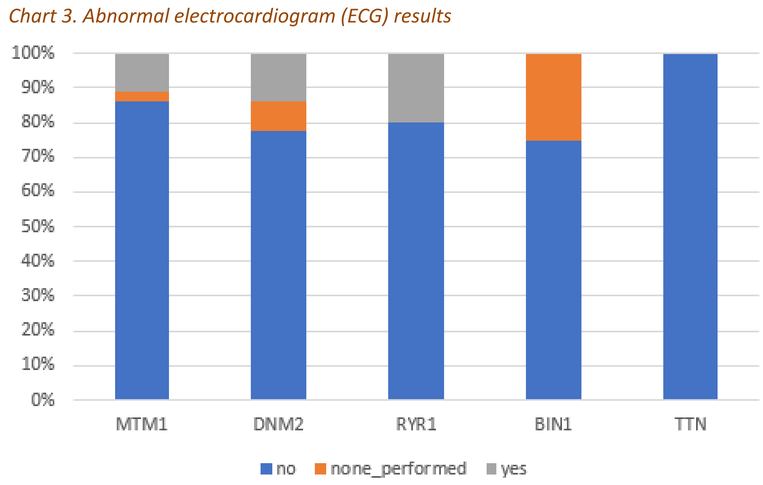


Chart 4. Reported eye movement

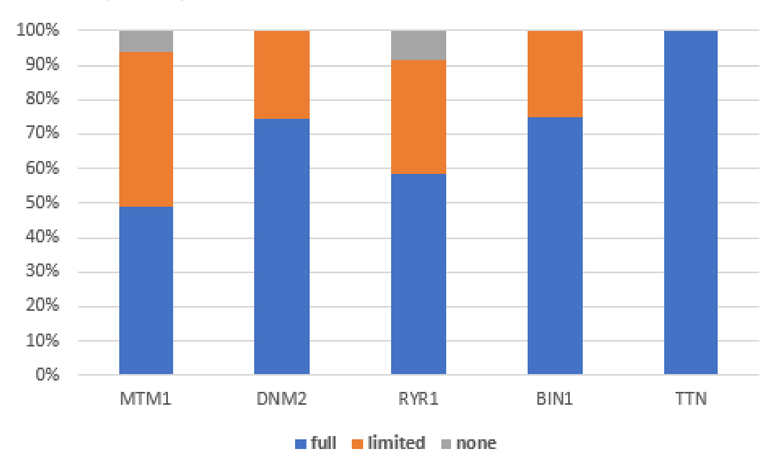


Chart 5. Consanguinity in family

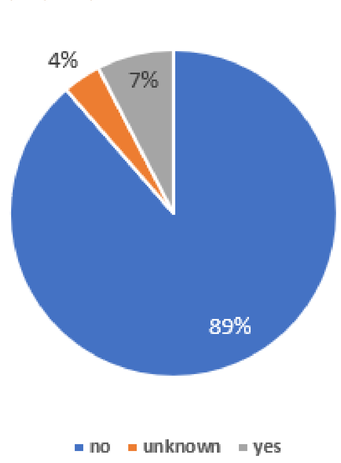
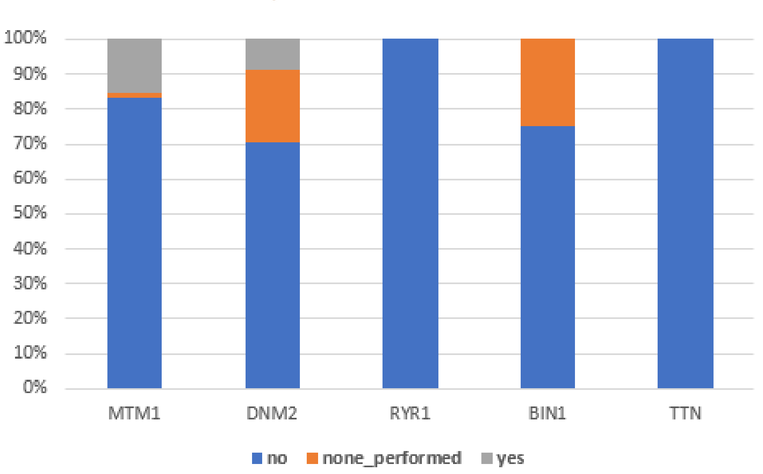


Chart 6. Abnormal echocardiogram results



SUMMARY AND FUTURE DEVELOPMENT

The MTM & CNM Patient Registry contains important data on a diverse and growing international cohort of individuals diagnosed with these conditions. It can support all areas of translational research including epidemiology, clinical trial planning and recruitment, outcome measure development, standards of care, and real-world data for regulatory decision-making. Continued investment will mean continued improvement in data quality, patient ascertainment, genetic confirmations, and overall value of the registry; unlocking its maximum potential as a prominent and fully utilised translational research tool for the entire disease community.

TREAT-NMD Neuromuscular Network *Member of the TREAT-NMD Neuromuscular Network*

Find out more: **www.mtmcnmregistry.org**
Enquiries: **mtmcnmregistry@newcastle.ac.uk**

Current funders:
myotubular trust FINDING STRENGTH
Muscular Dystrophy UK Fighting muscle-wasting conditions
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