Myotubular & Centronuclear Myopathy Patient Registry

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

The Myotubular and Centronuclear Myopathy Patient Registry

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

The John Walton Muscular Dystrophy Research Centre | Newcastle University | NHS The Newcastle upon Tyne Hospitals NHS Foundation Trust

WHAT IS THE REGISTRY?

The Myotubular and Centronuclear Myopathy (MTM and CNM) Patient Registry is an open-ended research database established in 2013, collecting important clinical and genetic information about people diagnosed with these conditions. Participants complete an online questionnaire and are asked to update this every 6 months.

The purpose of this registry is to:

- Help identify participants for clinical trials and other research studies.
- Understand the prevalence and geographical spread of these conditions.
- Collate information on the genetic causes, symptoms and severity of the conditions.
- Support existing research and encourage further research into the centronuclear myopathies.
- Inform health professionals to develop better standards of care for patients.
- Act as a communications link between the patient and research communities.

We welcome registrations of:

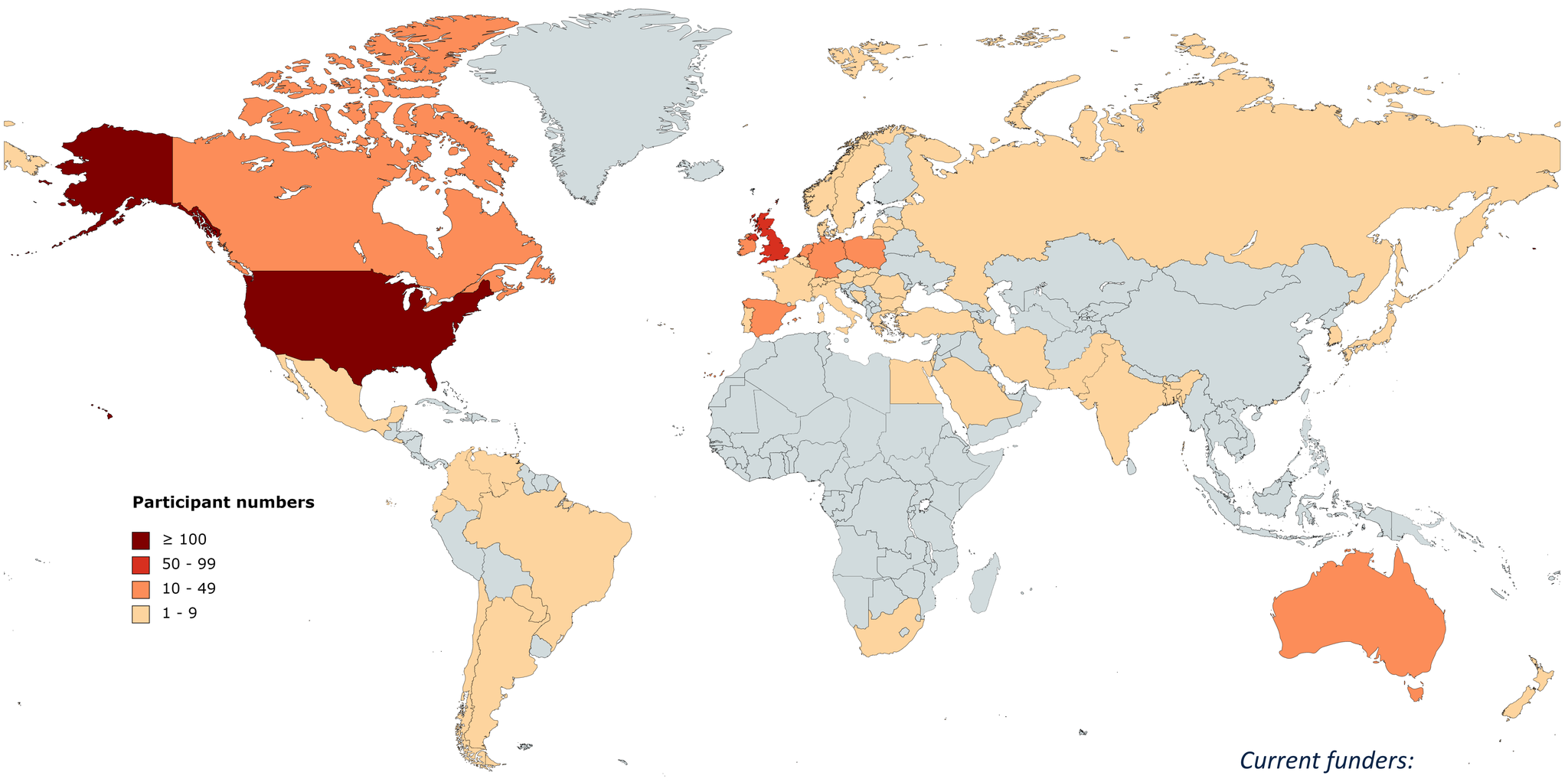
- ✓ Individuals with a diagnosis of MTM or CNM (preferably confirmed via genetic testing or muscle biopsy).
- ✓ Female carriers of X-linked MTM, especially those with myotubular myopathy symptoms.
- ✓ Any individual who is deceased, but had a confirmed diagnosis.

HOW IS REGISTRY DATA USED?

The information described above is collected and stored using a secure online portal, and then de-identified and made available to researchers or companies working in these conditions (if approved by the Registry Steering Committee).

Research we have supported so far includes:

- Incidence and prevalence of CNM
- Wearable devices and outcome measures
- Muscular symptoms in XLMTM carriers
- Motor function, ambulation and ventilation in XLMTM
- Fatigue in neuromuscular diseases
- Clinical features of MTM boys
- Causative genes in CNM
- Drug repurposing
- Experiences of conception, pregnancy and postpartum in neuromuscular diseases



INFORMATION COLLECTED

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

WHY JOIN THE REGISTRY?

Contribute to research

In such rare conditions as these, every piece of information is precious for researchers, health professionals, and drug regulators. The more we know about the number of people affected, and how they are affected, the more we can support research and therapy development.

Be visible for clinical trial planning and recruitment

When a company or clinical site is looking for people to take part in a clinical trial, they can ask us to send information about the trial to any registry participants who might be eligible. Registry data can also be used to help assess the feasibility and inform the planning of clinical trials.

Stay informed

Receive regular newsletters and ad-hoc news bulletins containing information about research, news and events relevant to your condition.

Help and signposting

Coordinated by a leading academic research centre at Newcastle University, and part of the international TREAT-NMD network, the Registry is well-placed to offer participants signposting or advice about many aspects of their condition or care.

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

Current funders: myotubular trust FINDING STRENGTH | Muscular Dystrophy UK Fighting muscle-wasting conditions | astellas GENE THERAPIES