EP.025 Myotubular and Centronuclear Myopathy Patient Registry: Accelerating the pace of research and treatment

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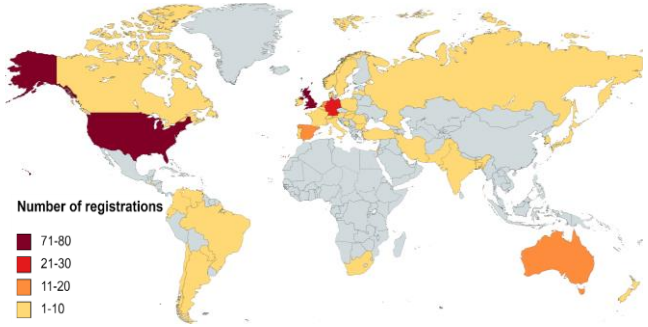
Introduction

Myotubular and other centronuclear myopathies are congenital neuromuscular conditions characterised by central nuclei on a muscle biopsy. The presumably most common form is the ultra-rare X-linked myotubular myopathy (XLMTM) with an estimated incidence of 1 in 50,000 male births. Mortality rates are not reliably ascertained but are believed to be 25-50% in the first year of life and ≈10% thereafter[1,2].

The Myotubular and Centronuclear Myopathy Patient Registry is a longitudinal, open-ended database collecting demographic, genetic and clinical data on affected individuals and female carriers of XLMTM from all over the world. Researchers or companies can:

- Request data to answer research questions.
- Ask the registry to inform eligible individuals about research studies and clinical trials.



Methods

Participants consent and provide data online against 12 domains (see right), and are asked to update this annually. Registrations are verified by registry staff following review of genetic or biopsy report and/or follow-up with clinician.

Questionnaires are available in six languages:

English German French Spanish Italian Polish

Registrations are accepted of:

- Children/adults diagnosed with MTM/CNM.
- Carrier females of XLMTM.
- Deceased individuals with a diagnosis.

- Genetic & biopsy information
- Clinical care
- Best and current motor function
- Wheelchair use
- Eye movement
- Scoliosis surgery
- Ventilation use
- Antibiotics
- Feeding tube use
- Heart function
- Family information
- Other registry participation

Question? Ask Jo now on Teams by scanning the QR code or visiting: tinyurl.com/askJoaquestion

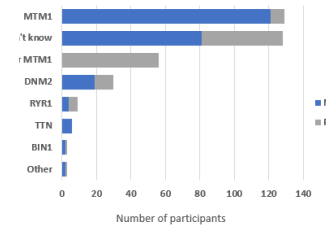
Next steps

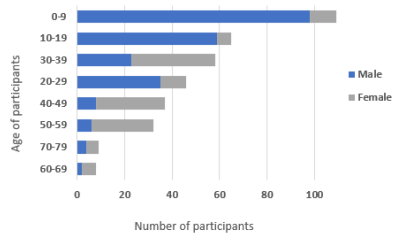
- Four additional languages
- Clinician-reported data
- Patient-reported outcome measures
- Publication of data

Results (June 2021)

364 participants:

- **330** living, **34** deceased
- **235** male, **129** female
- **183** living adults, **147** living children
- **46** countries





Conclusion and discussion

The Myotubular and Centronuclear Myopathy Patient Registry facilitates translational research by identifying research participants and providing important real-world data.

It contains important information on a diverse and growing international cohort of individuals affected by this group of rare and ultra-rare conditions. It aims to support all areas of translational research including epidemiology, clinical trial planning and recruitment, outcome measures, standards of care, and real-world data for regulatory decision-making.

Led by an academic group and an international independent steering committee, and funded by patient organisations and industry, the Registry showcases a collaborative community effort. Since 2014 it has provided data or recruitment support for multiple research studies. New grant funding from April 2021 to April 2023 has galvanised the project and enables exciting expansion and development to maximise the potential of this research study and scientific community resource.

Use case examples

Data Reports

- Incidence and prevalence of CNM
- Motor function, ambulation status and ventilation usage in XLMTM
- Causative genes in CNM
- Patient numbers against all data points, split by genetic confirmation
- Patient numbers against clinical trial eligibility criteria

Research Recruitment

- Experiences of conception, pregnancy and postpartum
- Phase 1/2 clinical trial recruitment
- Drug repurposing
- Muscular Symptoms of female XLMTM carriers
- Assessing fatigue in NMD
- Understanding dysphagia in NMD
- Wearable devices and outcome measures

References

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