

The Myotubular and Centronuclear Myopathy Patient Registry Newsletter



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Edition 1: December 2016

Welcome to the first bi-annual newsletter for the MTM and CNM Patient Registry. It's been just over 6 months since we launched the first phase of our new online registry at the 4th European Family Conference for Myotubular and Centronuclear Myopathy, on the 7th and 8th of May in Frankfurt, Germany.



Jo Bullivant
Registry Project Manager and Curator

One of the benefits of being part of this registry is that we will keep you updated with relevant news and developments, and this twice-yearly newsletter is one of the ways that we do this.

Please feel free to contact me with any questions or comments about the registry or the newsletter, or if you have any suggestions for the next edition.

www.mtmcnmregistry.org

The MTM and CMN Registry is an international, patient reported database of patients with myotubular or centronuclear myopathy. We also register female carriers of X-linked myotubular myopathy.

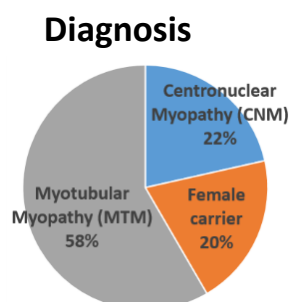
Registration is done through the website, and patients can create an online account so they can view or update their information at any time.

The registry aims to:

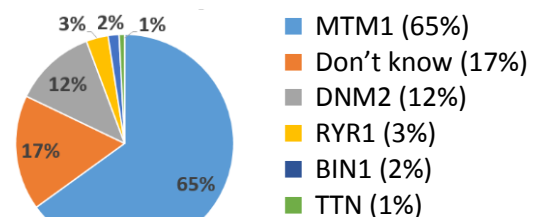
- **Help** identify patients for relevant clinical trials
- **Support** existing research projects with specific MTM and CNM data.
- **Encourage** further research into the conditions thanks to the existence and availability of data.
- **Provide** doctors with information on living with and managing these conditions, to help develop better standards of care.
- **Inform** participants about relevant news, research, studies, or clinical trials.

Who has registered? *

- ✓ 175 participants
- ✓ 121 male, 54 female
- ✓ Age ranging from 0 to 80 years
- ✓ Average age of patient: 21
- ✓ Average age of female carrier: 46
- ✓ From 26 countries



Affected genes



*We will never share identifiable or personal information about the patients on our registry.

Active or recruiting studies

These are exciting times for the MTM and CNM patient community, with promising progress being made in several different areas of research. New projects are started on a regular basis so it can be difficult to keep up. Here we have pulled together a list of currently active or recruiting studies, and their sponsors. If you would like to know more, you can use the links below to find the relevant contact details.

- [MTM Genetic Testing Study](#) (Observational study, Cure CMD)
- [Adverse Event Rates in Males with XLMTM](#) (Observational study, Cure CMD)
- [Respiratory Muscle Function in Untreated XLMTM](#) (Observational study, University of Florida)
- [Natural History and Functional Status of MTM Patients](#) (Observational study, Valerion Therapeutics/G n thon)
- [RECENSUS](#) and [INCEPTUS](#) (Observational study, Audentes Therapeutics)
- [Molecular Analysis of Congenital Myopathies](#) (Observational study, Boston Children's Hospital)

Data in the Registry

We collect a lot of information in our registry; here is a summary of the questions we ask:

1. Registration and demographics <ul style="list-style-type: none">Name and email addressDate of birth and genderAddress and phone numberOnline consent	2. Medical information <ul style="list-style-type: none">Genetic and biopsy reportsTesting/biopsy centre detailsDiagnosis and affected geneClinician details and diagnosisLast NM examination date
3. Motor function <ul style="list-style-type: none">Best and current motor functionWheelchair useEye movementScoliosis surgery	4. Respiratory function <ul style="list-style-type: none">Ventilation needs at birth and at presentVentilation type and frequencyAntibiotics for chest infections
5. Other functions <ul style="list-style-type: none">Feeding (gastric or nasal tube use)Heart (abnormal ECHO or ECG results)	6. Additional information <ul style="list-style-type: none">Family members with diagnosis or symptomsOther registries or studies joined

Meet our Steering Committee

The role of the registry steering committee is to make sure that the registry is well-governed, to ensure data quality and validation, and to oversee and approve any requests to use the registry or its data.



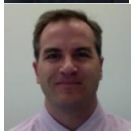
Hanns Lochmüller (Chair)

Professor of Experimental Myology and Consultant in Neuromuscular Genetics: Newcastle University, UK



Ana Buj Bello

Senior Scientist in Neuromuscular Disorders and Gene Therapy: INSERM, G n thon, France



James Dowling

Senior Scientist in Genetics and Genome Biology, and Staff Clinician in the Division of Neurology: Hospital for Sick Children, CA



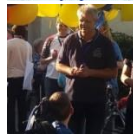
Anne Lennox

Chief Executive and Parent Trustee: Myotubular Trust, UK



Carsten B nnemann

Senior Investigator and Chief of Neuromuscular and Neurogenetic Disorders of Childhood: NINDS, USA



Aart den Hollander

Patient Representative: ZNM – Zusammen Stark! e.V., Netherlands



Heinz Jungbluth

Consultant in Paediatric Neurology and Senior Lecturer: Guy's and St Thomas' NHS Trust and King's College London, UK



Melanie Spring

Communications and Education Coordinator: Myotubular Trust, UK

Coming next year...

We have many exciting plans for 2017 to keep improving and expanding; including making the online registry available in different languages, accepting new registrations from female carriers and deceased patients, and moving patients from the old registry to the new online system. We will keep you all updated with our progress.

Useful resources

We understand that living with a rare disease can be overwhelming and at times confusing. Here we have listed a few resources that we hope will help you to feel informed:

- Are you protecting your personal data? Read our [Tips on Safe Data Sharing](#) to find out.
- Our friends at The MTM-CNM Family Connection have put together a [Basic Overview of the FDA Drug Approval Process](#) in America.
- Audentes Therapeutics have published a set of [Frequently Asked Questions](#) about AT132; their gene therapy product for X-linked myotubular myopathy.

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