

## MTM Brain MRI Study– Clinician Information

July 2024

Dear Clinician,

You are being contacted to make you aware of a new research study, the aim of which is to identify and characterize any abnormalities detected on MRI brain imaging in individuals with genetically confirmed **X-linked Myotubular Myopathy (XLMTM)**. We are currently recruiting patients and as you are a registered clinician with our registry, we have identified you as a potential contributor for this study.

### Who is involved?

SickKids Hospital, Toronto is leading this research study and has partnered with the Myotubular and Centronuclear Myopathy Patient Registry ([mtmcmregistry.org](http://mtmcmregistry.org)) at the John Walton Muscular Dystrophy Research Centre, Newcastle University, UK. Data will be entered and managed via the registry and de-identified MRI images and clinical data will be periodically transferred to the study team at SickKids for further analysis.

### How can you contribute?

1. Could you please identify patients at your institution who meet the study criteria
  - ✓ **genetically diagnosed XL MTM patients**
  - ✓ **previously received MRI neuroimaging.**
2. If any of your patients meet the above criteria, please encourage them to sign up through the registry ([mtmcmregistry.org](http://mtmcmregistry.org)) and provide them with the attached patient letter.
3. Once a patient provides consent for the study, the registry will then email asking you to complete the clinician questionnaire and upload the MRI files (and genetic report if not already provided by the patient). If you have any questions, you can contact me directly for support at [mtmcmregistry@newcastle.ac.uk](mailto:mtmcmregistry@newcastle.ac.uk)

Thank you very much for your involvement and anticipated support of this project!

Kindest Regards,



**Dr. Jim Dowling**, Principal Investigator, Sickkids Hospital, Toronto

**Dr. Lindsey Vogt, Kimberly Amburgey**, SickKids Hospital, Toronto

**Dr. Chiara Marini Bettolo**, MTM-CNM Registry Principal Investigator, Newcastle University, UK