

MTM Brain MRI Study Information

Patient Support Request - we need your help!

What is the study?

We want to inform you of a new research project being led by SickKids hospital, Toronto along with collaborators from other Canadian sites supported through data collection via the MTM-CNM international patient registry.

Currently there's not much known about rates of brain MRI changes in patients with X-linked Myotubular Myopathy (XLMTM). This study aims to collect previously undertaken brain MRI's for patients with XLMTM which will then be analysed to see if there are emerging patterns or changes.

Who can participate?

If you meet **all** the following criteria, you will be eligible to participate:

- ✓ Male patient (or parent/guardian of a male patient – living or deceased)
- ✓ Genetic confirmation of XLMTM
- ✓ Have previously had a brain MRI at any age (note: no new imaging will be obtained as part of this study)

How can you help us?

To be included as a study participant, you will need to be registered with the MTM and CNM Registry.

1) Currently Registered - please [log in](#) to the Registry and make sure all sections of the questionnaire are complete and up to date and follow the steps below:

- a) From **'Your Patient Data'**, select **'Other Functions'** and complete the small number of questions in the **'Brain & neurological conditions'** section. Click 'Save' at bottom of screen.
- b) From **'Your Patient Data'**, select your doctors name from the available list under **'Name your doctor'**. You can change a previous selection if you need to, or if your doctor is not shown on the list, contact the registry team mtmcmregistry@ncl.ac.uk and we will contact them and arrange to add them.
- c) Please speak to your doctor about the fact that you want to participate in this study, and we will contact them to advise on next steps.

2) Not currently registered – this is a great opportunity to join and it’s an easy process to [register with us](#). Please complete the patient details as far as possible, provide a genetic report and learn about the valuable work we do in supporting research to better understand the condition. Please ensure you complete the questions on the brain section so that you will be eligible to participate in this study.

What else will I need to do?

Once you’ve completed the actions above and spoken to your doctor, everything else will be done by the Registry in partnership with your doctor and the study team at SickKids.

Please be assured that all data obtained will comply with strict data protection protocols and MRI’s will be anonymised before being forwarded for analysis. If you have any questions or problems, please contact the Registry team at mtmcmregistry@ncl.ac.uk who will be happy to help you.

Your support is very much appreciated.

Kindest Regards,



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Dr. Lindsey Vogt - Paediatric Neurology Resident, SickKids, Toronto

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