PAGE 1: Medical Information

Knowing the precise details of an individual's mutation in an MTM or CNM gene will add to our understanding of these conditions and is likely to be important for developing treatments. As a carrier female of x-linked myotubular myopathy, you may or may not have had a genetic diagnosis yourself. However, a family member may have had a confirmed genetic diagnosis of x-linked myotubular myopathy.

If you have the genetic report yourself (or any other document that includes details of the genetic diagnosis) please upload a copy. Please also enter the name and contact details of the hospital, medical centre or geneticist centre where the genetic test was performed in the field below. In case anything is missing, we can easily obtain the correct document for you from them.

If you do not have the genetic report yourself (or the results of the genetic test are pending), please enter the name and contact details of the hospital, medical centre or geneticist where the test was performed in the field below. We will then contact them and ask for a copy of the report.

If a genetic test has not been performed yet, you may wish to contact your doctor, as knowing the details of the mutation is important if you have myotubular myopathy symptoms, and a treatment is required.

If a genetic mutation hasn't been confirmed in one of the MTM/CNM genes, you will need to have had a pathological diagnosis of myotubular/centronuclear myopathy via a **muscle biopsy**. Please note that it is quite likely that only those patients with a confirmed genetic mutation will be able to take part in clinical trials to assess potential treatments for these conditions. However those without a confirmed genetic diagnosis may be able to take part in research projects to find new genes for MTM or CNM.

For more help and information on how to be tested, speak with your doctor or geneticist.

Questions with a red asterisk* are mandatory

SECTION 1a: Genetic Report

Status of the genetic report *

- I have my genetic report and will upload a copy
- I do not have my genetic report myself but the genetic test results should be available
- The results of my genetic test are pending
- A genetic test has been performed on me but no mutation was found
- A genetic test was not performed on me
- A genetic test has not been performed on me, but a member of my family has a

confirmed genetic diagnosis of XLMTM.

A genetic test has been performed on me, but no mutation was found. But a member of my family has a confirmed genetic diagnosis of XLMTM.

Other (please specify)

If a genetic test was performed, please give the name and location of the testing hospital, medical centre, or laboratory: (free text box)

If you have the genetic report, please upload it here: (file upload option)

SECTION 1b: Muscle Biopsy

Has a muscle biopsy been performed? *
Yes / No / Don't know

If you answered yes, please tell us the name of the hospital where it was performed: (free text box)

If you answered yes, please tell us the location (city/country) of the hospital where it was performed: (free text box)

If you have the muscle biopsy report, please upload it here: (file upload option)

SECTION 1c: Clinician's Details

Please give the name and contact address of the *current* main specialist treating your neuromuscular condition * (this could be a family doctor, such as a GP): (free text box)

SECTION 1d: Neuromuscular Examination

Please provide the date of your last neuromuscular examination: * (free text box)

Month: Year Other:

Don't know

I've never had a neuromuscular appointment

PAGE 2: Other Functions

This page contains questions regarding your health.

Ventilation means breathing support from a mechanical ventilation device via a face or nose mask. Ventilatory support can be used either all day or for just a few hours.

Your **lung function** may have been done by spirometry reading (measurement of breath) or by sleep study.

Heart problems in myotubular myopathy and centronuclear myopathy patients are very rare, and it would be unusual for these tests to be 'abnormal'. However, it would be helpful if you could complete the questions.

You can only choose one of the given options. If none of the possibilities fits exactly, simply choose the one that is most appropriate.

Questions with a red asterisk* are mandatory.

SECTION 2a: Motor Function

Have you ever experienced weakness in walking or running? *

Don't know No Yes

Have you ever experienced difficulty with hills or steep slopes? *

Don't know No Yes

Have you ever experienced difficulty with stair climbing? *

Don't know No Yes

Have you ever experienced difficulty getting out of a sofa or off a low chair? *

Don't know No Yes

SECTION 2b: Respiratory Function

Have you ever required ventilation? *

Don't know No Yes

Have you ever had your lung function tested? *

Don't know No Yes

How many times have you required antibiotics for chest infections over the past 12 months? *

None required Between 1 and 3 times Between 4 and 6 times More than 6 times Don't know

SECTION 2c: Feeding Function

Have you ever had difficulty swallowing your food or drink? *

Don't know No Yes

SECTION 2d: Heart Function

Have you ever had an ABNORMAL echocardiogram (ECHO/Sonogram) result? *

Don't know No
Yes I have not had an echocardiogram

Have you ever had an ABNORMAL electrocardiogram (ECG) result? *

Don't know No
Yes I have not had an electrocardiogram

SECTION 2e: Other conditions

Not enough is currently known about how myotubular myopathy and other centronuclear myopathies might affect female carriers. We are interested in finding out whether female carriers on the registry have also been diagnosed with any of these more common conditions, in case there are any potential correlations or trends that we don't yet know about. Please note, this does not necessarily mean that we think female carriers might have a higher risk of developing these conditions.

Have you ever been diagnosed with, and treated for, any of the following? (Please tick all that apply)

- ☐ Anaemia
- ☐ Breast cancer
- ☐ Ovarian cancer
- ☐ Other cancer (please specify)
- ☐ Depression
- ☐ Heart disease (please give more details)
- ☐ Liver problems (please give more details)
- ☐ Osteoporosis
- ☐ None of the above

PAGE 3: Additional Information

Family information: Since myotubular myopathy and centronuclear myopathy are inherited conditions, it is important for us to know if there are any relatives who have similar symptoms or the same diagnosis.

Other Registries: Knowing whether you are registered elsewhere will help us to be accurate when we are estimating the prevalence of these conditions.

Newsletter preferences: Here you can tell us whether you would like to receive newsletters and general updates from us.

Questions with a red asterisk* are mandatory.

SECTION 3a: Family

Do you know of anybody else in your family who has been diagnosed with myotubular myopathy, centronuclear myopathy or similar symptoms? *

Don't know No Yes

To the best of your knowledge, has anyone in your family been married to a cousin or other blood relative? *

Don't know No Yes

SECTION 3b: Other Registries

Are your details registered with any other MTM or CNM-related registry or natural history study? *

Don't know

No, this is the only registry I have joined

Yes, I have joined another MTM and CNM related registry and/or natural history study

If you answered 'Yes', please tell us which ones:

Congenital Muscle Disease International Registry (CMDIR)
International Family Registry for Centronuclear and Myotubular Myopathies (Joshua Frase Foundation)
Don't know
Other

If you selected 'Other' please name the registry / registries: (free text box)

SECTION 3c: Newsletter Preferences

Would you like to receive general email communications relevant to Myotubular and Centronuclear Myopathy, such as newsletters, research results and standards of care? *
No / Yes

PAGE 4: Thank You For Your Registration

You have now completed the questionnaire.

Thank you for registering your details with the Myotubular and Centronuclear Myopathy Patient Registry.

Please make sure you have completed all the information you can. If there is any important information missing, we may contact you to ask about it.

Remember you can log back in at any time, using your email address and password, to complete or update your details.

You can now log out, or use the 'Modules' menu above to go back to different sections and add more information.

If you have any questions please contact the Registry Curator, Jo Bullivant, at mtmcnmregistry@treat-nmd.eu for help.

SECTION 4a: Any further comments?

If you have any feedback or comments on the registration process, please tell us here: (free text box)