MTM & CNM Patient Registry
Questionnaire for asymptomatic female carriers

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.

If you have the genetic report yourself (or any other document that includes details of the genetic diagnosis) please contact us at mtmcnmregistry@newcastle.ac.uk and we will send you a secure link where you can upload the document.

Please also enter the name and contact details of the hospital, medical centre or genetics 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 genetics centre 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, please contact your doctor, as knowing the details of the mutation is important.

Status of your genetic report

- ☐ I have my genetic report and will send / have sent 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 not been performed on me
- ☐ A genetic test has been performed on me but no mutation was found
- ☐ 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 and no mutation was found, but a member of my family has a confirmed genetic diagnosis of XLMTM.
- ☐ Not specified

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

[Free text field]

Has a muscle biopsy been performed?

- ☐ Yes
- ☐ No
- ☐ I don't know
- ☐ Not specified

If you answered yes, please tell us the name of the hospital where it was performed:

[Free text field]

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

[Free text field]

Please give the name and contact address of your current main doctor (this could be a family doctor such as a GP):

[Free text field]

Have you ever had a neuromuscular examination?

- ☐ Yes
- ☐ No
- ☐ I don't know
- ☐ Not specified

If yes, please provide the month and year of the most recent neuromuscular examination, if known

[Select month] [Select year]

MOTOR FUNCTION

Have you ever experienced weakness in walking or running?

- ☐ Yes
- ☐ No
- ☐ I don't know
- ☐ Not specified

Have you ever experienced difficulty with hills or steep slopes?

- ☐ Yes
- ☐ No
- ☐ I don't know
- ☐ Not specified

Have you ever experienced difficulty with stair climbing?

- ☐ Yes
- ☐ No
- ☐ I don't know
- ☐ Not specified

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

- ☐ Yes
- ☐ No
- ☐ I don't know
- ☐ Not specified

LIVER FUNCTION

Some (but not all) people with MTM and CNM have occasionally experienced abnormal liver function blood test results. Very rarely there are more severe liver related complications. In this section we hope to better understand the liver in myotubular and centronuclear myopathy, to help with day to day care of those with the conditions, and to improve future research.

Have you ever been diagnosed with a liver condition (cholestasis, non-alcoholic fatty liver, peliosis, Gilbert's syndrome, or other)?

- ○ Yes (please specify below)
- ○ No
- ○ I don't know
- ◉ Not specified

If yes, what is the liver condition?

When was it diagnosed?

As a newborn, were you treated for issues with high bilirubin or jaundice, including phototherapy perhaps received in the neonatal intensive care unit?

- ○ Yes
- ○ No
- ○ I don't know
- ◉ Not specified

If yes, did you require treatment for longer than 2 weeks?

- ○ Yes
- ○ No
- ○ I don't know
- ◉ Not specified

To the best of your knowledge, have you ever experienced any of the following possible liver-related symptoms?
Please select all that apply.

- [ ] Persistent, otherwise unexplained, itchy skin
- [ ] Jaundice (yellowing of the skin or eyes), not as a newborn
- [ ] Dark colour urine
- [ ] Pale stool colour
- [ ] Pain in abdomen
- [ ] None of the above
- [ ] I don't know

To the best of your knowledge, have any of the following lab test values been abnormal? Please select all that apply.

- [ ] Albumin
- [ ] ALP (alkaline phosphatase)
- [ ] ALT (alanine transaminase)
- [ ] AST (aspartate transaminase)
- [ ] Bilirubin (direct or indirect), not as a newborn
- [ ] GGT (gamma-glutamyl transferase)
- [ ] Prothrombin time (PT)/INR
- [ ] Serum bile acid test
- [ ] Liver function (specific test or enzyme unknown)
- [ ] None of the above
- [ ] I don't know

Have you ever been hospitalised as a result of your abnormal liver function lab values?

- ○ Yes (please provide details below)
- ○ No
- ○ I don't know
- ◉ Not specified

If yes, please provide details for each stay.

Type of hospitalisation

- ○ Planned (admission was scheduled in advance)
- ○ Emergency (admission was in response to an unexpected abnormal liver function lab value)
- ◉ Not specified

Approximate date

Add another hospitalisation

How often do you have blood drawn to test your liver function?

- ○ Never/not applicable

- ○ Yearly
- ○ Every 6 months
- ○ Other (please specify below)
- ○ I don't know
- ◉ Not specified

Other frequency

As part of standard screening for your muscle condition, you may sometimes undergo routine imaging scans. Have you ever had any of these imaging tests done on your liver?

Ultrasound

- ○ Yes
- ○ No
- ○ I don't know
- ◉ Not specified

If yes, when was the last ultrasound scan of your liver?

CT scan

- ○ Yes
- ○ No
- ○ I don't know
- ◉ Not specified

If yes, when was the last CT scan of your liver?

MRI

- ○ Yes
- ○ No
- ○ I don't know
- ◉ Not specified

If yes, when was the last MRI scan of your liver?

Fibroscan

- ○ Yes
- ○ No

- ○ I don’t know
- ◉ Not specified

If yes, when was the last fibroscan of your liver?

Other type of liver imaging

- ○ Yes
- ○ No
- ○ I don’t know
- ◉ Not specified

If yes, what type of liver imaging was performed?

When was the last liver scan of this type?

Have there been any abnormal findings reported based on your liver imaging?

- ○ Yes (please specifiy below)
- ○ No
- ○ I don’t know
- ◉ Not specified

If known, please describe the findings.

Who reviews your liver health?

- ☐ Hepatologist
- ☐ Gastroenterologist
- ☐ Primary care physican or GP
- ☐ Other (please specify below)
- ☐ No doctor reviews my liver health
- ☐ I don’t know

Other medical specialty

If a hepatologist reviews your liver health, what is their name and hospital or clinic? This information may help us plan future studies. We will not contact this doctor about you specifically.

How often do you have a follow-up review for your liver health?

- ○ Never/not applicable
- ○ Yearly
- ○ Every 6 months
- ○ Other (please specify below)
- ○ I don't know
- ◉ Not specified

Other frequency

Do you currently take any medications for liver issues?

- ○ Yes (please specify below)
- ○ No
- ○ I don't know
- ◉ Not specified

To the best of your knowledge, have you taken any medications for liver issues in the past?

- ○ Yes (please specify below)
- ○ No
- ○ I don't know
- ◉ Not specified

Please provide details of any liver medications you are taking currently or have taken in the past.
To add a further entry, click on the button "Add another medication".

Medication

☐ **I am currently taking this medication**

Start date

Stop date

Add another medication

Have you ever had a liver biopsy?

- ○ Yes
- ○ No
- ○ I don't know
- ◉ Not specified

If yes, when was this performed?

At what hospital?

How do liver issues currently affect your health?

- ○ Not at all: I do not have abnormal liver-related blood test and/or imaging results
- ○ Mildly: Blood tests and/or imaging have indicated some liver abnormalities, but I do not have symptoms that require medication to manage
- ○ Moderately: I have abnormal blood test and/or imaging results, along with liver-related symptoms that require medication to manage
- ○ Severely: I have liver-related symptoms that significantly impact my daily life (for example, requiring hospitalisation or frequent doctor visits)
- ◉ Not specified

Is there any other information you'd like to share regarding health issues that could be related to your liver?
Please note that this registry is only for research purposes. If you have any concerns, please speak to your doctor.

[Free text field]

OTHER FUNCTIONS

Have you ever required ventilation?
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.

- ☐ Yes
- ☐ No
- ☐ I don't know
- ☐ Not specified

Have you ever had your lung function tested?
Your lung function may have been done by spirometry reading (measurement of breath) or by sleep study.

- ☐ Yes
- ☐ No
- ☐ I don't know
- ☐ Not specified

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
- ☐ I don’t know
- ☐ Not specified

Have you ever had difficulty swallowing your food or drink?

- ☐ Yes
- ☐ No
- ☐ I don't know
- ☐ Not specified

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

- ☐ Yes
- ☐ No
- ☐ No echocardiogram has been done
- ☐ I don't know
- ☐ Not specified

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

- ☐ Yes
- ☐ No
- ☐ No electrocardiogram has been done
- ☐ I don't know
- ☐ Not specified

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

If requested above, please provide more information here:

[Free text field]

ADDITIONAL INFORMATION

Do you know of anybody else in your family who has been diagnosed with myotubular myopathy, centronuclear myopathy or has similar symptoms?
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. If so, please ask them to consider joining this registry, if they haven't already done so.

- ☐ Yes
- ☐ No
- ☐ I don't know
- ☐ Not specified

Are your biological parents related by blood as second cousins or closer?

- ☐ Yes
- ☐ No
- ☐ I don't know
- ☐ Not specified

Are your details registered with any other MTM or CNM-related registry or natural history study?
Knowing whether you are registered elsewhere will help us to be accurate when we are estimating the prevalence of these conditions.

- ☐ Yes, I have joined another MTM and CNM related registry and/or natural history study
- ☐ No, this is the only registry I have joined
- ☐ I don't know
- ☐ Not specified

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 field]

Have you ever taken part in a clinical trial?
Knowing whether you have ever taken part in a clinical trial will help us track the development and availability of new therapies and provide information to help with planning and feasibility studies for new clinical trials.

- ☐ Yes, I am currently taking part in a clinical trial
- ☐ Yes, I have previously taken part in a clinical trial
- ☐ No, I have never taken part in a clinical trial
- ☐ I don't know
- ☐ Not specified

If you answered Yes, please tell us the full name of the clinical trial.

[Free text field]

Please tell us how you heard about this registry.
This helps us to concentrate our efforts on the best ways to find other people who might want to join the registry.

- ☐ My doctor or other healthcare professional, e.g. physiotherapist, genetic counsellor)
- ☐ A patient support group (please tell us which one)
- ☐ Information included on a genetic test result
- ☐ A newsletter (please tell us which one)
- ☐ At a conference or other event (please tell us which one)
- ☐ Through social media, e.g., a Facebook group (please tell us which one)
- ☐ Word of mouth, from friends or family
- ☐ Other (please specify)

Details

[Free text field]

Would you like to receive general email communications relevant to Myotubular and Centronuclear Myopathy, such as newsletters, research results and standards of care?

- ☐ Yes
- ☐ No
- ☐ Not specified

If you have any feedback or comments on the registration process, please tell us here:

[Free text field]