MTM & CNM Patient Registry
Questionnaire for patients / parents of patients / symptomatic female carriers

YOUR DOCTOR

There are two parts to this registry; a questionnaire that you complete yourself, and a form that we ask your doctor to complete for you. To provide this information, your doctor will use their own login and will be able to see the information that you provide, but they will not be able to edit it.

Please select the name of the main doctor in charge of your care:

[Drop-down list]

If your doctor is not listed above, please tell us their details so we can invite them to take part. If you don't have a neuromuscular specialist, this could be a family doctor such as a GP.

Name of doctor

[Free text field]

The hospital or clinic where you see your doctor:

[Free text field]

Any contact details you have for the doctor (email, telephone number, etc.):

[Free text field]

Please declare your consent by reading and agreeing to the statement below:

I consent for my doctor named above to enter my medical data into the MTM & CNM Patient Registry. I consent to my named doctor having full read-only access to the data I have entered into the Registry.

[Tick field]

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 very important for an appropriate treatment.

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.

Status of the genetic report

- ☐ I have the genetic report and will send / have sent a copy
- ☐ I do not have the genetic report myself but the genetic test results should be available
- ☐ The results of the genetic test are pending
- ☐ A genetic test has not been performed
- ☐ A genetic test has been performed but no mutation was found

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]

Was the myotubular or centronuclear myopathy caused by a mutation in:

- ☐ MTM1 (x-linked myotubular myopathy)
- ☐ DNM2 (dynamin 2)
- ☐ BIN1 (amphysin II)
- ☐ RYR1 (skeletal muscle ryanodine receptor)
- ☐ TTN (titin)
- ☐ Don't know
- ☐ Other (please specify below)

Other mutation:

[Free text field]

Diagnosis, according to the specialist:

- ☐ Myotubular Myopathy (MTM)

- ☐ Centronuclear Myopathy (CNM)
- ☐ Other (please specify below)

Other diagnosis:

[Free text field]

Please provide the month and year of the most recent neuromuscular examination, if known:

[Select month] [Select year]

MOTOR FUNCTION

Motor function describes your ability to move your body. Sitting independently means that you can stay in a sitting position for several minutes, without being supported by another person or a stabilising device (such as a chair back, corset or brace).

Walking independently means without being supported by another person or stabilizing device (such as a walking frame, calipers or walking canes).

What is the best motor function ever achieved?

- ☐ Able to walk without support
- ☐ Able to walk with support
- ☐ Able to sit without support
- ☐ Never able to walk or sit independently
- ☐ I don't know
- ☐ Not specified

From and until what age was this best motor function achieved?

This motor function was achieved from the age of [] years and [] months up to the age of [] years and [] months (leave the last two fields blank if this is currently still the case).

What is the current motor function?

- ☐ Able to walk without support
- ☐ Able to walk with support
- ☐ Cannot walk but can sit independently (without support)
- ☐ Cannot walk or sit independently
- ☐ I don't know
- ☐ Not specified

From what age has this level of motor function been achieved?

This motor function has been achieved from the age of [] years and [] months.

Does [name] currently use a wheelchair?

- ☐ Always uses a wheelchair
- ☐ Sometimes uses a wheelchair, but able to walk short distances independently
- ☐ Always uses a wheelchair
- ☐ I don't know

If applicable:

Started to sometimes use a wheelchair for long distances at the age of [] years and [] months.

Started to always use a wheelchair to get around at the age of [] years and [] months.

How easily is [name] able to move his eyes?

This could be full range movement, or some limited movement where you can follow an object with your eyes, even if you can't move your head to look. If you are uncertain, please ask a medical professional such as your physiotherapist or doctor for advice.

- ☐ Full range movement of the eyes
- ☐ Some limited eye movement
- ☐ No eye movement
- ☐ I don't know
- ☐ Not specified

Has [name] had spinal surgery for scoliosis?

Some MTM or CNM patients suffer from weakness in their back muscles which results in a deformation or 'bending' of their spine called scoliosis. In order to stabilise the spine, they often have surgery done. In case such a surgery is planned, but has not yet been performed, please select 'No'

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

If you answered 'Yes', please provide the age when scoliosis surgery was performed.

Scoliosis surgery was performed at the age of [] years and [] months.

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.

Has [name] 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, was [name] 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 [name] require treatment for longer than 2 weeks?

- ○ Yes
- ○ No
- ○ I don't know

To the best of your knowledge, has [name] 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

Has [name] ever been hospitalised as a result of their 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 does [name] have blood drawn to test their 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. Has [name] ever had any of these imaging tests done on their liver?

Ultrasound

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

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

CT scan

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

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

MRI

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

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

Fibroscan

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

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

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

If known, please describe the findings.

Who reviews the patient's 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 the patient's 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 does [name] 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

Does [name] currently take any medications for liver issues?

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

To the best of your knowledge, has [name] 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 the patient is 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

Has [name] 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 the patient's 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 the liver?
Please note that this registry is only for research purposes. If you have any concerns, please speak to the patient's doctor.

[Free text field]

OTHER FUNCTIONS

Was ventilation required at birth?

Ventilation means breathing support from a mechanical ventilation device in the form of either non-invasive ventilation via a face or nose mask, or invasive ventilation via a tracheostomy (an operation to make an incision in the windpipe) or endotracheal tube (a breathing tube is inserted into the windpipe). Ventilatory support can be used either all day or for just a few hours.

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

What type of ventilation is currently used?

- ☐ Non-Invasive Ventilation (NIV) via a nose or face mask
- ☐ Invasive ventilation via endotracheal tube or tracheostomy
- ☐ No ventilation is currently needed
- ☐ Don't know

How many hours is ventilation currently used for?

- ☐ Full time (at least 16 hours per 24 hours)
- ☐ Part-time (less than 16 hours per 24 hours), while sleeping and while awake
- ☐ Part-time (less than 16 hours per 24 hours), but only while sleeping
- ☐ Ventilation is sometimes used as therapy, i.e. not regularly
- ☐ No ventilation is used
- ☐ I don't know
- ☐ Not specified

If ventilation is used, from what age has it been required?

Ventilation has been required from the age of [] years and [] months.

How many times have antibiotics for chest infections been required 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

Is a gastric or nasal tube currently used for feeding?

MTM/CNM patients sometimes have trouble eating and swallowing food orally (by mouth) and therefore have to be fed through a feeding tube. A gastric feeding tube (also called a G-tube or a Peg) is one that goes directly into the stomach through an incision in the tummy. A nasal feeding tube (also called nasogastric tube) is one that goes through the nose and down into the stomach.

- ☐ Yes, a feeding tube only

- ☐ Yes, a feeding tube with some oral feeding
- ☐ No
- ☐ I don't know
- ☐ Not specified

Has [name] ever had an ABNORMAL echocardiogram (ECHO/Sonogram) result?

Many neuromuscular patients are routinely required to have regular echocardiograms (ECHO / Sonogram) and electrocardiograms (ECG). 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 still be helpful if you could complete the questions.

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

Has [name] ever had an ABNORMAL electrocardiogram (ECG) result?

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

ADDITIONAL INFORMATION

Do you know of anybody else in [name]'s 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 with 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 [name]'s biological parents related by blood as second cousins or closer?

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

Are [name]'s 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
- ☐ No
- ☐ 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)
- ☐ I don't know
- ☐ Other

If you selected 'Other' please name the registry / registries:

[Free text field]

Has [name] ever taken part in a clinical trial?

This 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, currently participating in a clinical trial
- ☐ Yes, previously participated in a clinical trial
- ☐ No, never participated 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]

DOCTOR QUESTIONNAIRE

The individual named above has provided consent for you to enter their medical data into this registry. They will be able to view (but not edit) any information you provide, and you will be able to view (but not edit) the information they have provided. If you see any information provided by them that you believe is incorrect, please let the Registry Curator know at mtmcnmregistry@newcastle.ac.uk.

DIAGNOSIS

Clinical diagnosis

- ☐ Myotubular Myopathy
- ☐ Female carrier of XLMTM
- ☐ Centronuclear Myopathy
- ☐ Other (specify – free text)
- ☐ Unknown

Has the diagnosis above been genetically confirmed?

- ☐ Yes
- ☐ No
- ☐ Unknown

If 'Yes':

Upload genetic report (if available and if not provided already by participant.)

Enter genetic mutation using format 'Gene c. ..... p.' (For example RYR1 c.476G>A p.Arg177Cys)

If 'No':

Is there a biopsy report available that is supportive of a CNM diagnosis?

- ☐ Yes and I will upload
- ☐ Yes but I don't have it

- ☐ No
- ☐ Unknown

If you have it, please upload a copy

Clinical features

Best motor function ever achieved (use definitions from WHO Motor Milestones):

- ☐ Walking alone (without assistance)
- ☐ Walking with assistance
- ☐ Sitting without support
- ☐ Unable to walk or sit independently
- ☐ Unknown

At what age was this level of motor function achieved?

- ☐ [Numerical field] (YY)
- ☐ Unknown

Are they still able to achieve this?

- ☐ Yes (*skip to 'Was ventilation required at birth?')*
- ☐ No
- ☐ Unknown

If No:

At what age was it lost?

- ☐ [Numerical field] (YY)
- ☐ Unknown

Current motor function (use definitions from WHO Motor Milestones):

- ☐ Walking alone (without assistance)
- ☐ Walking with assistance
- ☐ Sitting without support
- ☐ Unable to walk or sit independently
- ☐ Unknown

At what age was this level of motor function achieved?

- ☐ [Numerical field] (YY)
- ☐ Unknown

Was ventilation required at birth?

- ☐ Yes
- ☐ No
- ☐ Unknown

Is ventilation currently used?

- ☐ Yes
- ☐ No
- ☐ Unknown

If Yes

From what age has ventilation been used?

- ☐ [Numerical field] (YY)
- ☐ Unknown

Current ventilation type

- ☐ IV (Invasive ventilation)
- ☐ NIV (Non-Invasive Ventilation) via BiPAP
- ☐ NIV (Non-Invasive Ventilation) via CPAP
- ☐ Unknown

Current ventilation frequency

- ☐ Full-time (≥16 hours per 24 hours)
- ☐ Part-time (<16 hours per 24 hours), both awake and sleeping
- ☐ Part-time (<16 hours per 24 hours), only while sleeping
- ☐ Unknown

Thank you for your contribution to the registry. We will contact you in 6 months and ask you to either update this record or confirm that nothing has changed.

If you have any questions or comments please contact the Registry Curator at mtmcnmregistry@newcastle.ac.uk.