MTM & CNM Patient Registry

Questionnaire for registering a deceased family member

GENERAL INFORMATION

This questionnaire contains questions regarding the deceased patient. We realise that this may be difficult for you, but please try to answer all the questions as fully as you can. This information is likely to be important for developing treatments and finding out the best way to care for other affected people.

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

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 click on the following link to upload the document:

Upload document (link opens in new window)

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 from them.

If you do not have the patient's 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 mutation hasn't been confirmed in one of the MTM/CNM genes, the patient would have needed to have had a pathological diagnosis of myotubular/centronuclear myopathy via a muscle biopsy.

Status of the patient’s genetic report

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

Was a muscle biopsy 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 their form of 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)
- I don't know
- Other (please specify below)

Other mutation:

[Free text field]

Please give the name and contact address of the main specialist or specialists who treated the patient's neuromuscular condition

[Free text field]

What was their diagnosis, according to their doctor?

- Myotubular Myopathy (MTM)
- Centronuclear Myopathy (CNM)
- Other (please specify below)
- Not specified

Other diagnosis:

[Free text field]

MOTOR FUNCTION

Motor function describes a person’s ability to move his or her body. Sitting independently means that he or she could stay in a sitting position for several minutes, without being supported by another person or a stabilizing device (such as a chair back, corset or brace). Walking independently means walking without being supported by another person or stabilizing device (such as a walking frame calipers or walking canes).

What was the best motor function the patient 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 level of best motor function achieved?

This motor function was achieved from the age of [] years and [] months up to the age of [] years and [] months.

Did they use a wheelchair?

- They always used a wheelchair
- They sometimes used a wheelchair, but were able to walk short distances independently
- They did not use a wheelchair
- I don't know
- Not specified

If applicable:

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

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

How easily were they able to move their eyes?

Eye movement could have been either full range movement, or some limited eye movement where they could follow an object with their eyes, even if they couldn’t move their head to look.

- They had full range movement of the eyes
- They had some limited eye movement
- They had no eye movement
- I don't know
- Not specified

Did they have spinal surgery for scoliosis? *

Some MTM/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.

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

If you answered 'yes' above, please provide the age when scoliosis surgery was performed

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

OTHER FUNCTIONS

Did the patient require ventilation at birth?

Ventilation means breathing support from a mechanical ventilation device in the form of either non-invasive ventilation (NIV 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 could have been used either all day or for just a few hours.

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

Did they ever regularly need to use ventilation?

- Yes, 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
- They sometimes used ventilation as a therapy, i.e. not regularly
- No, they did not use ventilation
- I don’t know
- Not specified

Did they ever use non-invasive ventilation (NIV) via a nose or face mask?

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

If they required non-invasive ventilation (NIV) via a mask, from and until what age did they use this type of ventilation?

Please try to give your answer in years and months, for example "from birth, until 3 months".

[Free text field]

Did they ever use invasive ventilation via a tracheostomy or endotracheal tube?

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

If they required invasive ventilation (via endotracheal tube or tracheostomy), from and until what age did they use this type of ventilation?

Please try to give your answer in years and months, for example "from birth, until 3 months".

[Free text field]

How many times were they given antibiotics for chest infections over a 12 month period (or less if they were under a year old)?

- None required
- Between 1 and 3 times
- Between 4 and 6 times
- More than 6 times
- I don't know
- Not specified

Did they use a gastric or nasal tube 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

Did they ever have 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 be helpful if you could complete the questions.

- Yes
- No
- They did not have an echocardiogram
- I don't know
- Not specified

Did they ever have an ABNORMAL electrocardiogram (ECG) result?

- Yes
- No
- They did not have an electrocardiogram
- I don't know
- Not specified

ADDITIONAL INFORMATION

Do you know of anybody else in the patient'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 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 his biological parents related by blood as second cousins or closer?

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

Were this patient's details ever registered with any other registry or natural history study?

Knowing whether the patient was 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]

Did this patient ever take part in a clinical trial?

Knowing whether this patient ever took 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
- No
- 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 take part in the registry.

- Our 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]