PARTICIPANT QUESTIONNAIRE

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.

Questions with a red asterisk * are mandatory.

Section 1a: Patient details

1. **Please tell us your relationship to this patient** *

   The patient was my child

   The patient was not my child, but I was his/her guardian

   The patient was my parent

   The patient was my sibling

   The patient was my cousin, aunt, etc (please specify – free text box)

2. **What was their date of death?** * **(DD/MM/YYYY)**

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 patient's 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 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 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 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.

Questions with a red asterisk * are mandatory

Genetic Report

3. **Status of genetic report** *

I have their genetic report and will upload 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

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

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

Muscle Biopsy

6. **Was a muscle biopsy performed?** *

Yes / No / Don't know

7. **If you answered 'Yes', please tell us the name of the hospital where it was performed:** (free text box)

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

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

Genetic Mutation

10. **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)

Don't know

Other (please specify)

Clinician's Details

11. **Please give the name and contact address of the main specialist or specialists who treated the patient's neuromuscular condition** * (free text box)

Diagnosis

12. **What was their diagnosis, according to their doctor?** *

Myotubular Myopathy (MTM)

Centronuclear Myopathy (CNM)

Other (please specify – free text box)

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).

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.

Spinal surgery: 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.

Questions with a red asterisk * are mandatory

Motor Function

13. What was their best motor function ever achieved? *

Walking without support

Walking with support

Sitting without support

Never able to walk or sit independently

Don't know

14. From and until what age was this level of best motor function achieved?

Please give your answer in months and years, for example: "From 2 years 1 month, until 16 years 2 months"

15. Did they use a wheelchair? *

They did not use a wheelchair

They sometimes used a wheelchair, but they were able to walk short distances independently

They always used a wheelchair

Don't know

16. From and until what age was this level of motor function achieved? (free text box)

Please give your answer in months and years, for example: "From 2 years, 0 months, until 4 years, 11 months"

17. How easily were they able to move their eyes? *

They had full range movement of the eyes

They had some limited eye

movement They had no eye

movement

Don't know

18. **Did they have spinal surgery for scoliosis?** *

No Yes

19. **If you answered 'yes' above, please provide the age when scoliosis surgery was performed** (free text box)

Please give your answer in months and years, for example "3 years and 2 months".

Other Functions

Respiratory function

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 wind-pipe) or endotracheal tube (a breathing tube is inserted into the wind-pipe). Ventilatory support could have been used either all day or for just a few hours.

Feeding function

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.

Heart function

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.

Questions with a red asterisk * are mandatory

Respiratory Function

20. **Did they require ventilation at birth?** *

Don't know No Yes

21. Did they ever regularly need to use ventilation? *

No, they did not use ventilation

They sometimes used it as a therapy, i.e. not regularly

Yes, for several hours a day and/or at night

Yes, full time, i.e. 24 hours a day

Don't know

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

No, they did not use non-invasive ventilation Yes, they used non-invasive ventilation (NIV) Don't know

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

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

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

No, they did not use invasive ventilation Yes, they did use invasive ventilation Don't know

25. 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".

Antibiotics

26. 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 Don't know

Feeding Function

27. Did they use a gastric or nasal tube for feeding? *

No

Yes, a feeding tube with some oral feeding Yes, a feeding tube only

Don't know

Heart Function

28. Did they ever have an ABNORMAL echocardiogram (ECHO/Sonogram) result? *

Don't know No

Yes They did not have an echocardiogram

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

Don't know No

Yes They did not have an electrocardiogram

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 the patient was registered elsewhere will help us to be accurate when we are estimating the prevalence of these conditions.

Clinical Trials: 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.

How you found us: This helps us to concentrate our efforts on the best ways to find other people who might want to take part in the registry.

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

Family information

30. **Do you know of anybody else in the patient's family who has been diagnosed with myotubular myopathy, centronuclear myopathy or similar symptoms?** *

    Don't know No Yes

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

    Don't know No Yes

Other registries

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

    Don't know No Yes

33. **If you answered "Yes", please provide the names of the registries and/or natural history studies this patient is registered with:**

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

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

Clinical trials

35. **Did this patient ever take part in a clinical trial?**

Yes
No
I don't know

36. If you answered Yes, please tell us the name of the clinical trial.
(Free text)

How you found us

37. Please tell us how you heard about this 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)

Newsletter Preferences

38. 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

Thank you for your registration

You have now completed the questionnaire.

Thank you for registering this patient's 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.

If you haven't completed all the details yet, you can log back in at any time, using your email address and password.

You can now log out, or go back to different sections and add more information.

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

Any further comments?

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