Myotubular and Centronuclear Myopathy Patient Registry

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Data source | Human | Disease registry

Administrative details

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Data source ID

45942

Data source acronym

MTM & CNM Patient Registry

Data holder

John Walton Muscular Dystrophy Research Centre, Newcastle University

Data source type

Disease registry

Main financial support

Funding from industry or contract research

Funds from patients organisations, charity and foundations

Care setting

Other

Data source qualification

If the data source has successfully undergone a formal qualification process (e.g., from the EMA, ISO or other certifications), this should be described.

No

Data source website

https://mtmcnmregistry.org

Contact details

Julie Bohill mtmcnmregistry@newcastle.ac.uk

Main

mtmcnmregistry@newcastle.ac.uk

Jess Page Jess.Page@newcastle.ac.uk

Alternate

Jess.Page@newcastle.ac.uk

Data source regions and languages

Data source countries

United Kingdom

Data source languages

English

Data source establishment

Data source established

26/03/2013

Data source time span

First collection: 26/03/2013

The date when data started to be collected or extracted.

Publications

Data source publications

Publications not yet available - Please see link to landing page on clinicaltrials.gov

Data elements collected

The data source contains the following information

Disease information

Does the data source collect information with a focus on a specific disease? This might be a patient registry or other similar initiatives.

Yes

Disease details (other)

Myotubular myopathy, centronuclear myopathy, female carriers of x-linked myotubular myopathy

Rare diseases

Are rare diseases captured? In the European Union a rare disease is one that affects no more than 5 people in 10,000.

Yes

Pregnancy and/or neonates

Does the data source collect information on pregnant women and/or neonatal subpopulation (under 28 days of age)?

Yes

Hospital admission and/or discharge

No

ICU admission

Is information on intensive care unit admission available?

No

Cause of death

Not Captured

Prescriptions of medicines

Not Captured

Dispensing of medicines

Not Captured

Advanced therapy medicinal products (ATMP)

Is information on advanced therapy medicinal products included? A medicinal product for human use that is either a gene therapy medicinal product, a somatic cell therapy product or a tissue engineered products as defined in Regulation (EC) No 1394/2007 [Reg (EC) No 1394/2007 Art 1(1)].

No

Contraception

Is information on the use of any type of contraception (oral, injectable, devices etc.) available?

No

Indication for use

Does the data source capture information on the therapeutic indication for the use of medicinal products?

Not Captured

Medical devices

Is information on medicinal devices (e.g., pens, syringes, inhalers) available?

No

Administration of vaccines

No

Procedures

Does the data source capture information on procedures (e.g., diagnostic tests, therapeutic, surgical interventions)?

Captured

Procedures vocabulary

Other

Procedures vocabulary, other

Bespoke multiple-choice questionnaire

Healthcare provider

Is information on the person providing healthcare (e.g., physician, pharmacist, specialist) available? The healthcare provider refers to individual health professionals or a health facility organisation licensed to provide health care diagnosis and treatment services including medication, surgery and medical devices.

Yes

Clinical measurements

Is information on clinical measurements (e.g., BMI, blood pressure, height) available?

No

Genetic data

Are data related to genotyping, genome sequencing available?

Captured

Genetic data vocabulary

HGNC

Biomarker data

Does the data source capture biomarker information? The term “biomarker” refers to a broad subcategory of medical signs (objective indications of medical state observed from outside the patient), which can be measured accurately and reproducibly. For example, haematological assays, infectious disease markers or metabolomic biomarkers.

Not Captured

Patient-reported outcomes

Is information on patient-reported outcomes (e.g., quality of life) available?

No

Patient-generated data

Is patient-generated information (e.g., from wearable devices) available?

Yes

Units of healthcare utilisation

Are units of healthcare utilisation (e.g., number of visits to GP per year, number of hospital days) available or can they be derived? Units of healthcare utilisation refer to the quantification of the use of services for the purpose of preventing or curing health problems.

No

Unique identifier for persons

Are patients uniquely identified in the data source?

Yes

Diagnostic codes

Captured

Diagnosis / medical event vocabulary

Not coded (Free text)

Medicinal product information

Not Captured

Quality of life measurements

Not Captured

Lifestyle factors

Not Captured

Sociodemographic information

Captured

Sociodemographic information collected

Age

Country of origin

Gender

Quantitative descriptors

Population Qualitative Data

Population age groups

Paediatric Population (< 18 years)

Preterm newborn infants (0 – 27 days)

Term newborn infants (0 – 27 days)

Infants and toddlers (28 days – 23 months)

Children (2 to < 12 years)

Adolescents (12 to < 18 years)

Adults (18 to < 46 years)

Adults (46 to < 65 years)

Elderly (≥ 65 years)

Adults (65 to < 75 years)

Adults (75 to < 85 years)

Adults (85 years and over)

Estimated percentage of the population covered by the data source in the catchment area

Exact prevalence and incidence of the centronuclear myopathies are unknown. An estimated incidence of 17 per million births for XLMTM (the main subtype of CNM) corresponds to an estimated prevalence of 4,679, but current estimates do not fully capture the true picture. There are 450 participants in the Registry, of whom 153 reported a diagnosis of XLMTM, corresponding to 3% of the

conservative estimates described above.

The age range of current living registry participants is 0-86 years.

Description of the population covered by the data source in the catchment area whose data are not collected (e.g., people who are registered only for private care)

Any affected individuals worldwide who have either (a) never heard of the registry or (b) decided not to participate

Family linkage

Family linkage available in the data source permanently or can be created on an ad hoc basis

Ad hoc

Population

Population size

450

Active population size

354

Data flows and management

Access and validation

Governance details

Documents or webpages that describe the overall governance of the data source and processes and procedures for data capture and management, data quality check and validation results (governing data access or utilisation for research purposes).
https://www.mtmcnmregistry.org

Biospecimen access

Are biospecimens available in the data source (e.g., tissue samples)?

No

Access to subject details

Can individual patients/practitioners/practices included in the data source be contacted?

Yes

Description of data collection

Online registry platform. Users join and maintain their data through a secure online portal.

Event triggering registration

Event triggering registration of a person in the data source

Other

Event triggering registration of a person in the data source, other

Patient-initiated (voluntary) registration.

Event triggering de-registration of a person in the data source

Loss to follow up

Other

Event triggering de-registration of a person in the data source, other

Patient asks to be removed from the registry or provides a medical report which confirms they do not have a diagnosis of MTM or CNM.

Event triggering creation of a record in the data source

Participants receive an automated email 6 months after their last log-in, prompting them to log in again and update their questionnaire.

Data source linkage

Linkage

Is the data source described created by the linkage of other data sources (prelinked data source) and/or can the data source be linked to other data source on an ad-hoc basis?

Yes

Linkage description, pre-linked

In development

Linkage description, possible linkage

In development

Data management specifications that apply for the data source

Informed consent for use of data for research

Other

Possibility of data validation

Can validity of the data in the data source be verified (e.g., access to original medical charts)?

No

Data source preservation

Are records preserved in the data source indefinitely?

Yes

Approval for publication

Is an approval needed for publishing the results of a study using the data source?

Yes

Informed consent, other

There is a steering committee to evaluate requests for data access. Participants consent to the use of their data for purposes approved by the steering committee.

Common Data Model (CDM) mapping

CDM mapping

Has the data source been converted (ETL-ed) to a common data model?

No