The United Kingdom Spinal Muscular Atrophy Patient Registry

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Data source | Human | Disease registry

Administrative details

Administrative details

Data source ID

1000000029

Data source acronym

UK SMA 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

Hospital outpatient care

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

UK SMA Patient Registry

Contact details

Registry Curator smaregistry@newcastle.ac.uk

Main

smaregistry@newcastle.ac.uk

Data source regions and languages

Data source countries

Ireland

United Kingdom

Data source languages

English

Data source establishment

Data source established

07/07/2008

Data source time span

First collection: 07/07/2008

The date when data started to be collected or extracted.

Publications

Data source publications

Prevalence, incidence and carrier frequency of 5q–linked spinal muscular atrophy – a literature review

Real-World Data on Access to Standards of Care for People With Spinal Muscular Atrophy in the UK

A multi-source approach to determine SMA incidence and research ready population

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

Spinal muscular atrophy

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

Yes

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

Not coded (Free text)

Other

Procedures vocabulary, other

Participants are able to select from a specific list of procedures. If their procedure is not listed, they can report it as free text.

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?

Yes

Genetic data

Are data related to genotyping, genome sequencing available?

Captured

Genetic data vocabulary

Other

Genetic data vocabulary, other

Curator is able to select from a specific list of SMA-related genetic mutations. If the specific genetic mutation is not listed, it can be reported as free text.

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.

Captured

Biomarker data vocabulary

Other

Biomarker vocabulary, other

Free text

Patient-reported outcomes

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

Yes

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)

Other

Diagnosis / medical event vocabulary, other

Participants are able to select their diagnosis from a specific list. If their diagnosis is not listed, they can report it as free text.

Medicinal product information

Not Captured

Quality of life measurements

Captured

Quality of life measurements vocabulary

EQ5D

other

Quality of life measurements, other

Patient-reported global impression of change; SMA independence scale (SMAIS)

Lifestyle factors

Not Captured

Sociodemographic information

Not Captured

Quantitative descriptors

Population Qualitative Data

Population age groups

All

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)

Adult and elderly population (≥18 years)

Adults (18 to < 65 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

Studies estimate that between one and two people in every 100,000 worldwide are living with spinal muscular atrophy (SMA) at any one time. The incidence of SMA is estimated as one in every 10,000 babies worldwide are born with a type of SMA. There are 669 participants in the Registry which very approximately represents 46% of the SMA population in the UK and Ireland.

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 nationwide 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

669

Active population size

421

Population by age group

Age group	Population size	Active population size
Paediatric Population (< 18 years)	245	151
Infants and toddlers (28 days – 23 months)	5	5
Children (2 to < 12 years)	147	101
Adolescents (12 to < 18 years)	93	45
Adults (18 to < 46 years)	264	163
Adults (46 to < 65 years)	110	82
Elderly (≥ 65 years)	50	25
Adults (65 to < 75 years)	26	16

Age group	Population size	Active population size
Adults (75 to < 85 years)	21	9
Adults (85 years and over)	3	0

Median observation time

Median time (years) between first and last available records for unique individuals captured in the data source

1.00

Median time (years) between first and last available records for unique active individuals (alive and currently registered) capt

4.00

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).

Legal Notice, Privacy Notice and Data Protection Statement

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. Participants 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

New registration received (patient-initiated). Once registered, participants receive invitations every six months to review and update their data.

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

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

Event triggering creation of a record in the data source

Patient-initiated (voluntary) registration.

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

The UK SMA Patient Registry was not created by the linkage of other data sources.

Linkage description, possible linkage

Consent is collected for data linkage between the UK SMA Patient Registry and SMA REACH UK (paediatric) and Adult SMA REACH clinical databases. Data linkage is achieved through a manual process. All three databases are independent and their IT platforms are not connected. The purpose of the linkage is to link patient-reported outcome measures (PROMs) from the patient registry with clinical data from SMA REACH.

Linked data sources

Pre linked

Is the data source described created by the linkage of other data sources?

No

Data source, other

Adult SMA REACH

Linkage strategy

Other

Linkage variable

A single variable is used, referred to as the SMA REACH reference number (e.g. SN1234). The single variable is created by the SMA REACH data sources. If a patient is participating in either of these studies, they will be allocated an SN number which they can report to the UK SMA Patient Registry.

Linkage completeness

Approximately 50% of participants in the UK SMA Patient Registry have reported an SMA REACH reference number.

Pre linked

Is the data source described created by the linkage of other data sources?

No

Data source, other

SMA REACH UK

Linkage strategy

Other

Linkage variable

A single variable is used, referred to as the SMA REACH reference number (e.g. SN1234). The single variable is created by the SMA REACH data sources. If a patient is participating in either of these studies, they will be allocated an SN number which they can report to the UK SMA Patient Registry.

Linkage completeness

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Data management specifications that apply for the data source

Data source refresh

Every 6 months

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