Longitudinal Data Collection from Patients with Spinal Muscular Atrophy (SMArtCARE)

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Data source | Human | Disease registry

Administrative details

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PURI

https://redirect.ema.europa.eu/resource/42196

Data source ID

42196

Data source acronym

SMArtCARE

Data holder

University Medical Center Freiburg

Data source type

Disease registry

Main financial support

Funding from industry or contract research

Care setting

Hospital inpatient care

Hospital outpatient care

Primary care – GP, community pharmacist level

Primary care – specialist level (e.g. paediatricians)

Secondary care – specialist level (ambulatory)

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

SMArtCARE Registry for Spinal Muscular Atrophy

Contact details

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Data source regions and languages

Data source countries

Austria

Germany

Switzerland

Data source languages

English

German

Data source establishment

Data source established

15/06/2017

Data source time span

First collection: 15/06/2017

The date when data started to be collected or extracted.

Publications

Data source publications

SMArtCARE - A platform to collect real-life outcome data of patients with spinal muscular atrophy(Pechmann A et al., Orphanet Journal of Rare Diseases, 2019)

Zur Gentherapie der Spinalen Muskelatrophie mit Onasemnogene Abeparvovec. Stellungnahme der Gesellschaft für Neuropädiatrie

Development and Pilot Test of the Registry Evaluation and Quality Standards Tool: An Information Technology-Based Tool to Support and Review Registries

Efficacy and safety of gene therapy with onasemnogene abeparvovec in children with spinal muscular atrophy in the D-A-CH-region: a population-based

observational study

Clinical Effectiveness of Newborn Screening for Spinal Muscular Atrophy: A Nonrandomized Controlled Trial.

5qSMA: standardised retrospective natural history assessment in 268 patients with four copies of SMN2. 5qSMA: standardised retrospective natural history assessment in 268 patients with four copies of SMN2.

Long-term efficacy and safety of nusinersen in adults with 5q spinal muscular atrophy: a prospective European multinational observational study.

Improvements in Walking Distance during Nusinersen Treatment - A Prospective 3-year SMArtCARE Registry Study.

Improved upper limb function in non-ambulanSMA type 2 and 3 during nusinersen treatment: a prospective 3-years SMArtCARE registry study.

Effect of nusinersen on motor, respiratory and bulbar function in early-onset spinal muscular atrophy.

Studies

List of studies that have been conducted using the data source

A Prospective, Long-Term Registry of Patients with a Diagnosis of Spinal Muscular Atrophy (SMA) - (RESTORE)

A Prospective, Observational, Post-Authorisation Efficacy Study to Assess Long-term Effectiveness of Risdiplam in Patients with Genetically Confirmed 5q SMA

Spinraza (nusinersen) SMA Pregnancy Exposure Study Within Existing SMA Registries

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

Captured

Cause of death vocabulary

ICD-10

Prescriptions of medicines

Captured

Dispensing of medicines

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)].

Yes

Contraception

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

Yes

Indication for use

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

Captured

Indication vocabulary

Other

Indication vocabulary, other

Only one indication encoded. We can derive subtypes from other variables and code them on orphacode (or ordo).

Medical devices

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

Yes

Administration of vaccines

Yes

Procedures

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

Captured

Procedures vocabulary

Not coded (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

Not coded (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

Not coded (free text)

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.

Yes

Unique identifier for persons

Are patients uniquely identified in the data source?

Yes

Diagnostic codes

Captured

Diagnosis / medical event vocabulary

ICD-10

MedDRA

Medicinal product information

Captured

Medicinal product information collected

Active ingredient(s)

Brand name

Dose

Route of administration

Medicinal product vocabulary

ATC

Quality of life measurements

Captured

Quality of life measurements vocabulary

Not coded (Free text)

Lifestyle factors

Not Captured

Sociodemographic information

Captured

Sociodemographic information collected

Age

Gender

Quantitative descriptors

Population Qualitative Data

Population age groups

All

Paediatric Population (< 18 years)

Adults (18 to < 65 years)

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

This assessment cannot be made.

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)

Nation-wide

Family linkage

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

Ad hoc

Population

Population size

2129

Population by age group

Age group	Population size
Paediatric Population (< 18 years)	1248
Adult and elderly population (≥18 years)	881

Median observation time

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

3.80

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).

http://www.smartcare.de

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

After site initiation, documenting users apply for a user account for the EDC-system Insight by OpenApp ltd., users login in with username and password into EDC-system to document Enrolment, Baseline and following visits until End of Data Collection; PRO included in Medical Assessment

Event triggering registration

Event triggering registration of a person in the data source

Disease diagnosis

Other

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

Filled in patient informed consent

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

Death

Emigration

Loss to follow up

Event triggering creation of a record in the data source

Specialist encounter, medicinal product dispensing, AE

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, possible linkage

Double pseudonymisation of data and creation of unique identifiers for FAIRification process. We use the "FAIR in an box" tool to produce data in RDF format and provide them on a graph database. We use a FAIR Data point for linkage of metadata. Once spider pseudonymization service is set up, we will use spider's probablistic (firstname, lastname, dob) pseudonym generation for record linkage.

Linked data sources

Pre linked

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

No

Data source, other

EURO-NMD (https://ern-euro-nmd.eu/)

Linkage strategy

Probabilistic

Linkage variable

We will use spider's generated pseudonyms in the future.

Linkage completeness

It depends on the required variables; many variables in EURO-NMD are more applicable to other NMD registries, so not all the variables can be linked to SMArtCARE.

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)?

Yes

Data source preservation

Are records preserved in the data source indefinitely?

No

Data source preservation length (years)

10 years

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 committee to evaluate requests for data access

Common Data Model (CDM) mapping

CDM mapping

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

Yes

CDM Mappings

CDM name (other)

Set of common data elements for rare diseases registration

Data source ETL status

Not ETL-ed

Data source ETL specifications (link)

https://eu-rd-platform.jrc.ec.europa.eu/sites/default/files/CDS/EU_RD_Platform_...