A registry-based cohort study of Spinal Muscular Atrophy (SMA) disease to describe the natural history of SMA, the evolution of SMA care management and disease progression considering new disease modifying therapies (DMTs).

First published: 27/01/2023
Last updated: 04/05/2024

Study | Ongoing

Administrative details

PURI
https://redirect.ema.europa.eu/resource/50477

EU PAS number
EUPAS50476

Study ID
50477

DARWIN EU® study
No

Study countries
Austria
Belgium
Czechia
Germany
Ireland
Slovakia
Spain

Sweden
United Kingdom

Study description
To investigate SMA patients' course of disease and standards of care delivery over time in multiple European countries:
Objective 1: To describe, by SMA type, the natural history of SMA (the disease and its progression) in the UNTREATED cohort and the TREATED cohort also stratified by DMT, including patients characteristics, disease progression based on motor function assessment as well as respiratory, nutritional and skeletal deformities, post-diagnostic outcomes of interest and serious adverse events of special interest.
Objective 2: To describe by SMA type the evolution of diagnosis methods and of medicinal and non-medicinal treatment over time, including adoption of DMTs in the "ALL" cohort and the DMTs patterns.

Study status
Ongoing

Research institution and networks

Institutions

Aetion
Spain
First published: 24/11/2022
Last updated 16/07/2024

Institution | ENCePP partner | Other

- Swedish National Registry for Neuromuscular disorder (Neuromuskulära sjukdomar i Sverige - NMiS), Sweden
- Belgian Neuromuscular Diseases Registry (BNMDR), Belgium
- REaDY, Czech Republic & Slovakia
- Registro Nacional de Pacientes de la Fundación Atrofia Muscular Espinal (FundAME), Spain

- DMD- und SMA-Patientenregister für Deutschland und Österreich), Germany & Austria
- UK SMA Patient Registry, UK & Ireland

Networks

Translational Research in Europe - Assessment and Treatment of Neuromuscular Diseases (TREAT-NMD)

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Network

Contact details

Study institution contact
Elizabeth Garry

Study contact

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Primary lead investigator
Nicolas Deltour

Primary lead investigator

Study timelines

Date when funding contract was signed
Planned:
06/04/2022
Actual:
06/04/2022

Study start date
Planned:
05/12/2022
Actual:
27/01/2023

Data analysis start date
Planned:
27/01/2023
Actual:
15/03/2023

Date of interim report, if expected
Actual:
11/10/2023

Date of final study report
Planned:
02/06/2023

Sources of funding

- EMA
- EU institutional research programme

Study protocol

Protocol_EMA_SMA Version 1.3 Signed March 3 2023 (1).pdf(1.51 MB)

Regulatory

Was the study required by a regulatory body?
Yes

Is the study required by a Risk Management Plan (RMP)?
Not applicable

Methodological aspects

Study type

Study type list

Study type:
Non-interventional study

Scope of the study:
Disease epidemiology
Drug utilisation

Main study objective:
To describe, by SMA type, the natural history of SMA in the UNTREATED cohort and the TREATED cohort also stratified by DMT, incl patients characteristics, disease progression
To describe by SMA type the evolution of diagnosis methods and of medicinal and non-medicinal treatment over time, including adoption of DMTs in the “ALL” cohort and the DMTs patterns.

Study Design

Non-interventional study design
Cohort

Study drug and medical condition

Medical condition to be studied
Spinal muscular atrophy

Population studied

Age groups
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 number of subjects
2188

Study design details

Data analysis plan
Mainly descriptive study

Documents

Study report
SIGNED-SMA EMA - Report - FINAL- April 5 2024 + Appendix D-E-F.docx_.pdf(9.26 MB)

Study, other information
Objective 0 (Preliminary) Results 2023_10_10.xlsx_.pdf(589.55 KB)
Objective 1 (Natural History) Results 2024_1_26.pdf(1.07 MB)
Objective 2 (Healthcare) Results 2023_10_10.xlsx - All.pdf(224.62 KB)
Supplementary Results Objective 1 (Natural History) 2024_1_26.pdf(8.15 MB)
Supplementary Results Objective 2 (Healthcare) 2023_11_9.xlsx_.pdf(508.97 KB)

Data management

Data sources

Data source(s)
Translational Research in Europe - Assessment and Treatment of Neuromuscular Diseases

Data sources (types)
Disease registry

Use of a Common Data Model (CDM)

CDM mapping
No

Data quality specifications

Check conformance
Unknown

Check completeness
Unknown

Check stability
Unknown

Check logical consistency
Unknown

Data characterisation

Data characterisation conducted
No