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

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 institutions and networks

Institutions

Aetion

Spain

First published: 24/11/2022

Last updated: 16/07/2024

Institution | Other | ENCePP partner

- 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

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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:

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

ENCePP Seal

The use of the ENCePP Seal has been discontinued since February 2025. The ENCePP Seal fields are retained in the display mode for transparency but are no longer maintained.

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