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





### 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

### 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.

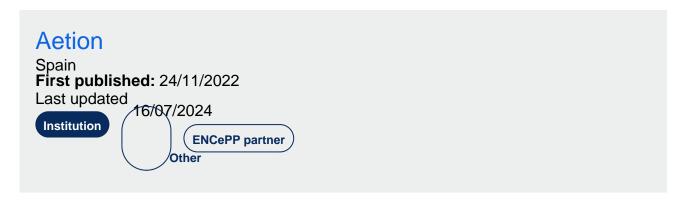
Objective 2: To describe by SMA type the evolution of diagnosis methods and of medicinal and non-medicinal treatment over time, including adoption of DMTs in the "ALL" cohort and the DMTs patterns.

### Study status

Ongoing

### Research institution and networks

### Institutions



- 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)

First published: 01/02/2024

Last updated 01/02/2024

Network

### Contact details

Study institution contact

Elizabeth Garry

Study contact

liz.garry@aetion.com

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 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