

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

**First published:** 27/08/2021

**Last updated:** 03/09/2024

Study

Ongoing

## Administrative details

### EU PAS number

EUPAS41853

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### Study ID

41854

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### DARWIN EU® study

No


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### Study countries

 Argentina

 Chile

 Greece

 Ireland

-  Japan
  -  Korea, Democratic People's Republic of
  -  Poland
  -  Portugal
  -  Romania
  -  Russian Federation
  -  Taiwan
  -  United States
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## Study description

This prospective observational registry will assess long-term outcomes of patients with a diagnosis of SMA, including long term safety and effectiveness of OAV-101.

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## Study status

Ongoing

## Research institutions and networks

### Institutions

[Novartis Pharmaceuticals](#)

**First published:** 01/02/2024

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**Institution**

### Networks

iSMAC, French SMA Registry, SMArtCARE,  
CuidAME, Cure SMA

## Contact details

### Study institution contact

Novartis Clinical Disclosure Officer  
[trialandresults.registries@novartis.com](mailto:trialandresults.registries@novartis.com)

Study contact

[trialandresults.registries@novartis.com](mailto:trialandresults.registries@novartis.com)

### Primary lead investigator

Novartis Clinical Disclosure Officer

Primary lead investigator

## Study timelines

### Date when funding contract was signed

Planned: 01/10/2017

Actual: 12/01/2018

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### Study start date

Planned: 25/09/2018

Actual: 25/09/2018

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**Data analysis start date**

Planned: 25/09/2018

Actual: 31/12/2019

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**Date of interim report, if expected**

Planned: 12/12/2019

Actual: 02/07/2020

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**Date of final study report**

Planned: 28/10/2038

## Sources of funding

- Pharmaceutical company and other private sector

## More details on funding

Novartis

## Study protocol

[PRO-1300 RESTORE AVXS-101-RG-001\\_Redacted.pdf](#) (1.75 MB)

[PMA1234\\_RESTORE Protocol Amendment 3.0\\_V4.0 final\\_28Sep2023 - signed\\_Redacted.pdf](#) (784.1 KB)

## Regulatory

**Was the study required by a regulatory body?**

Yes

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## Is the study required by a Risk Management Plan (RMP)?

EU RMP category 2 (specific obligation of marketing authorisation)

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### Regulatory procedure number

AVXS-101-RG-001

## Other study registration identification numbers and links

AVXS-101-RG-001, NCT04174157, COAV101A12001

[Link to ClinicalTrials.gov](#)

## Methodological aspects

### Study type

#### Study type list

##### Study type:

Non-interventional study

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##### Scope of the study:

Other

Safety study (incl. comparative)

##### If 'other', further details on the scope of the study

Safety and efficacy study of OAV-101 in the real-world setting

##### Main study objective:

This registry will assess long-term outcomes of patients with a diagnosis of SMA. It will also characterize and assess long-term safety and effectiveness of OAV-101 in the real-world setting.

## Study Design

### **Non-interventional study design**

Other

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### **Non-interventional study design, other**

Prospective, multi center, multinational, non-interventional observational registry of patients diagnosed with SMA.

## Study drug and medical condition

### **Medical condition to be studied**

Spinal muscular atrophy

## Population studied

### **Age groups**

- Term newborn infants (0 - 27 days)
  - Infants and toddlers (28 days - 23 months)
  - Children (2 to < 12 years)
  - Adolescents (12 to < 18 years)
  - Adults (18 to < 46 years)
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### **Estimated number of subjects**

## Study design details

### Outcomes

To assess the effectiveness of treatments for SMA, characterize motor performance, assess the long-term safety of OAV-101, characterize risk of hepatotoxicity, thrombocytopenia, thrombotic microangiopathy, cardiac AEs and sensory abnormalities suggestive of ganglionopathy in SMA patients treated with OAV-101, assess ventilation-free survival and overall survival of all patients with SMA. To assess healthcare utilization, caregiver burden and patient functional independence. To characterize the natural history/epidemiology of patients with less than 4 copies of the SMN2 gene. To characterize the use of systemic glucocorticosteroids and other systemic immunosuppressive medication used to help manage the humoral immune response to the AAV9 vector.

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### Data analysis plan

Data will be analyzed per the statistical analysis plan (SAP). The analysis populations will consist of all patients enrolled. The primary analysis will be to summarize outcomes by the therapy a patient was on at the time of enrollment. Descriptive statistics will be presented for the primary analysis. No formal a priori hypothesis testing will be performed. Continuous variables will be summarized using the number of observations, mean, 95% confidence interval (CI) for the mean, standard deviation (SD), standard error (SE), median, minimum, and maximum. Categorical data will be summarized using counts and percentages. Incidence rates (per person-years) and 95% CIs of AEs will be calculated. Survival will be presented using Kaplan-Meier methods. Further data analysis may be undertaken to meet specific regulatory requests.

## 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)

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

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### Data sources (types)

[Disease registry](#)

[Electronic healthcare records \(EHR\)](#)

[Other](#)

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### Data sources (types), other

Prospective patient-based data collection

## Use of a Common Data Model (CDM)

### CDM mapping

No

## Data quality specifications

### Check conformance

Unknown

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**Check completeness**

Unknown

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**Check stability**

Unknown

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**Check logical consistency**

Unknown

## Data characterisation

**Data characterisation conducted**

No